Institute for Prospective Technological Studies

joint research centre
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Joint Research Centre (DG JRC)

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Report EUR 20516 EN

FOREWORD

Genetic testing is used to identify alterations in the DNA sequence that correlate with a disease or higher risk to develop a disease. This type of tests can be used for diagnosis before any symptoms of disease are recognisable and to determine the personal risk for certain multifactorial diseases. Thus, the results of genetic testing can have far reaching effects on an individual's life.

The European Commission's Joint Research Centre (JRC) , in particular its Institute for Prospective Technological Studies (IPTS), has been active in the field of genetic testing services since 1999. Within the IPTS' *Futures* project (1999) quality standards of genetic testing services was one of the topics discussed. In July 2000, IPTS organised a workshop, including all major stakeholders, where the need to better ensure the quality of genetic testing services at EU level was stressed.

The JRC received, in October 2001, a request from DG RTD to carry out a prospective study on the technical needs and options for the quality assurance and harmonisation of genetic testing services in Europe. In response to this request, IPTS and IRMM (Institute for Reference Materials and Measurements) began work on a project that aims to the identification and assessment of the means to guarantee that genetic testing services provide the same high quality within the enlarged EU.

In March 2002, a workshop with international experts was organised by IPTS to discuss the current situation and to identify lines of work towards achieving those goals. This workshop has been the starting point of a recently launched study, to be done in collaboration with the ESTO Network, to assess different harmonisation scenarios. During this workshop, experts and JRC agreed on the need to obtain updated "field" data on the situation of genetic testing services in each of the Member States. It was deemed essential, in order to start the study, to obtain and present background information for each Member State on

- the number of tests and conditions tested per year,
- the main centres providing the services and their regional distribution,
- ownership (public vs. private) and type of centre activity (health care, research or teaching),
- the framework for accreditation of services and personnel,
- the referral system in place, specially for rare diseases,
- data handling policies (informed consent and privacy issues),

and other data that would provide an accurate picture of the current situation on which to build our research. This has been attempted several times and reports exist, but the information is fragmented and/or out of date.

This is why, in order to provide an example of the type of information that would be required, IPTS asked the Andalusian Agency for Health Technology Assessment (AETSA) to provide a pilot report on the situation of genetic services in one Member State (Spain). This report will be given to the consortium formed in the ESTO to illustrate what ideally would be needed for the whole, enlarged EU.

The pilot survey on the current situation and practices of genetic services for hereditary diseases in Spain was commissioned to AETSA because of their ample experience in this type of studies. They had carried similar surveys at national level in other health technologies and they had full support from the Spanish Society for Human Genetics, which was essential in this case.

The survey was revealing as to the constantly increasing use of genetic testing in the EU, the current needs, and timely emergence of the JRC activity in the field.

Emilio Rodriguez-Cerezo
Life Sciences Co-ordinator, IPTS
Seville, October 2002

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EXECUTIVE SUMMARY

This report presents the results of a survey on the current situation and practices of genetic testing services for hereditary diseases in Spain, including tests available, characteristics of centres, patient referral pathways and accreditation processes.

The study targeted molecular DNA testing for diagnosis of hereditary diseases, excluding other types of diagnostic tests, such as cytogenetics, biochemical testing, or pharmacogenetics. The survey was carried out at the beginning of 2002.

A total of 53 centres performing genetic testing for hereditary diseases in Spain were identified, of which 42 are public and 11 private. The majority of laboratories belong to a hospital, and there is at least one centre performing genetic testing in each of the 17 autonomous regions of Spain. However, some regions as Catalonia, Madrid, Aragon, Basque Country or Galicia have higher numbers of centres per capita than the rest.

Due to the decentralised organisation of the Spanish health systems, a majority of centres are receiving patients from other regions or even countries, for at least one given hereditary disease, without any formal organisation. An official accreditation system is not in place, but the Spanish Society for Human Genetics provides an accreditation system for professionals. Some Spanish centres are participating in quality control schemes developed by the European Molecular Genetics Quality Network (EMQN) or the Cystic Fibrosis European Network. At least two centres are in the process of getting the ISO 9000 – 9002 accreditation.

According to survey results, 214 genetic conditions are currently being tested in Spain. The conditions most frequently tested are those with a higher frequency in general population (hemochromatosis, coagulation disorders, fragile X, cystic fibrosis or haematologic cancers) and they are performed in several centres and regions.

In Spain, there are no specific regulations or guidelines on molecular genetic testing, neither at state nor at regional level. Specific committees on genetic testing do not exist. Referral of patients for genetic testing is nearly exclusive of hospitals and specialised care, without any established standard protocol. It can be performed for clinical reasons or as a part of a research protocol. According to the survey, informed consent is requested by 90% of centres but there is no harmonised procedure. Usually, the physician requesting the test gets informed consent from the patient, although there could be important gaps in this process.

More than 95% of the tests are financed by public institutions (hospitals or universities) from their global budgets, without specific earmarked money. Usually centres and labs perform genetic testing only for a small number of conditions, and in many cases the test protocols used are home-brew. Payment for the tests performed in other centres, either in Spain or abroad, could be in some cases cumbersome due to the lack of established mechanisms, particularly for rare diseases.

Overall, the results from this pilot study show that the system lacks clear organisation and there is no formalised quality assurance scheme in place, although some centres participate in European schemes. "Non-official" reference centres for rare diseases appear and disappear depending on research trends. At the same time, the response to the survey from all centres was very positive and most of them acknowledged the need and willingness to join a European level quality assurance harmonisation effort. There is also an increasing trend in the number of types of hereditary diseases for which tests are being offered, which is a direct consequence of continuous research closely connected to practice.

The survey presented a picture of the situation of Spain that seems to be concordant with the situation in many Member States.

1. Introduction

Many hereditary diseases are known to be caused by a change in the DNA code of a single gene, also called monogenic diseases. The number of monogenic diseases described is continuously increasing and in the year 2000, the data bank of Online Mendelian Inheritance in Men (OMIM)[1] included about 1500 clinical disorders for which mutations were listed. This number increases as more is known about the human genome. Currently, the laboratories included in the European Directory of DNA Diagnostic Laboratories (EDDNAL)[2] perform genetic diagnostic tests for 580 genetic conditions.

Apart from monogenic diseases, there are other diseases in which hereditary factors play a role, but which are determined multifactorially, i.e. their development usually involves several genes and also environmental factors. Among these disorders are hereditary types of cancer, susceptibility to thrombosis, asthma, hypertension, diabetes, Alzheimer disease, atherosclerosis or others. These multifactorial diseases not only concern rare mutations, but also combinations of frequently occurring variations (polymorphisms). The relationship between a mutation and the onset of the disease is often less clear in these cases. The likelihood of a disease occurring as a result of a mutation varies considerably. In the case of factor V Leiden mutation in heterozygotes, for example, the likelihood of venous thrombosis is four to nine times greater, with a risk of 0.1 percent per year of developing symptoms. On the other hand, some genetic risk factors are very powerful, as in the case of BRCA mutations with a 60-85 percent risk of developing breast or ovarian cancer in carriers

An important number of the hereditary conditions that can be detected by means of genetic testing are "rare diseases", that is those diseases with a prevalence of less than 5 per 10,000 in the population. With Decision No 1295/1999/EC, the European Parliament and the Council of the European Union of 29 April 1999 adopted a programme of Community action on rare diseases within the framework for action in public health (1999 to 2003).

In spite of their low prevalence, these diseases are responsible for conditions that imply an important detriment for those who suffer them and their families. Also these diseases have an important effect on the demand for health and social services. At the same time, provision and quality assurance of genetic testing for rare hereditary diseases pose distinct problems. Given that research into genetic mutation is often complex, while laboratories performing genetic testing of the most common diseases exist in most European countries, only a few laboratories are in the position to supply an appropriate test for certain rare diseases.

Genetic testing differs from other medical testing in that it can be predictive for future onsets of (severe) disorders and that it can also be relevant for the patient's relatives. The information obtained from genetic tests is invariable, as it refers to a DNA sequence, therefore tests are carried out only once. Thus, the results of genetic testing

[1] http://www.ncbi.nlm.nih.gov/OMIM/
[2] http://www.eddnal.com/

can have far reaching effects on an individual's life, which renders quality assurance of genetic testing services an issue of utmost importance.

The estimated incidence or prevalence of some hereditary diseases for which tests are available is presented in Table 1. For instance hemochromatosis, the most frequent hereditary genetic disease, affects about one in two hundred people, but it has been estimated that one in eight people of Northern European descent are carriers of the disease. Population screening for these conditions could have important consequences and proposals in that direction should be rigorously evaluated.

Table 1. Estimated incidence or prevalence of some hereditary diseases

Disease	Estimated incidence or prevalence (Source)
Becker muscular dystrophy	Birth incidence: 1 in 18,500 live born males (EMQN)
Cystic fibrosis	Incidence: 1 in 2,500 live births (Gelehrter 1998)
Duchenne muscular dystrophy	Birth incidence: 1 in 4,000 live born males (EMQN)
Familial breast cancer	Incidence: 4 to 8 in 1,000 women (EMQN)
Familial adenomatous polyposis coli	Prevalence: 1 in 8,000 to 1 in 13,500 live births (EMQN)
Fragile-X syndrome	Prevalence: 1 in 5000 males (EMQN). 1 in 1039 males (Pembrey 2001)
Friedreich ataxia	Prevalence: 1 to 2 in 100,000 (EMQN)
Haemophilia A	Prevalence: 1 in 5,000 males (EMQN)
Haemophilia B	Prevalence: 1 in 25,000 males (EMQN)
Hemochromatosis	Prevalence: 5 in 1,000 people (Bulaj 2000)
Hereditary motor and sensory neuropathies	Prevalence: 10 to 40 in 10,000 (EMQN)
Hereditary non-polyposis coli	0.5% to 13% of all the colorectal cancer at the population level (Percesepe 2001)
Huntington disease	Incidence: 0.65 per 100,000 per year (McCusker 2000); Prevalence: 5.38 in 100,000 (Burguera 1997)
Mitochondrial disorders	Incidence of mitochondrial encephalomyopathies in children <6 years of age 1 in 11,000. Leigh's syndrome 1 in f 32,000, and both Alper's syndrome and infantile mitochondrial myopathy with cytochrome C oxidase deficiency 1 in 51,000. The point prevalence (as of January 1, 1999) of mitochondrial encephalo-myopathies in children under 16 years of age is 1 in 21,000. (Darin 2001)
Myotonic dystrophy	Incidence: 1 in 8,000 (EMQN)
Prader Willi / Angelman syndromes	Incidence: 1 in 15,000 to 1 in 20,000 (EMQN)
Spinal muscular atrophy	Birth incidence: 1 in 10,000 (EMQN)
Spinocerebellar ataxias (other than Friedreich ataxia)	Prevalence: 1 in 100,000 (EMQN)
Y-chromosome microdeletions	Prevalence: 7 in 100 infertile men (EMQN)

*EMQN: http://www.emqn.org/bpguidelines.htm (accessed 2002-03-23)

1.1 Technical issues

Genetic testing is a field with technical particularities because it changes following the rapid developments around the recently opened genomics era. The vast amount of information generated every year by genomics is translated into new genetic tests for diseases. These tests have little time to move from the research status to the quality status required for a routine clinical analysis. Also, since the technology and the knowledge are so new, very often the research labs are doing the tests (because the typical clinical labs are not used or do not master the techniques). Research labs are not accustomed to the quality control needed in routine clinical laboratories

Genetic testing implies accurate observation of one or more areas of a gene to check whether a specific alteration can be observed. Some diseases are linked to one alteration located in a well known specific area of one gene, a *hot spot*, a situation which simplifies the diagnosis of the genetic alteration. That is the case for some diseases like fragile-X disease, Steiner disease or some ataxias. In frequent genetic diseases such as hemochromatosis or cystic fibrosis there are two or three hot spots in which the predominant mutations can be easily diagnosed. In other cases, like breast cancer mutations, there are no hot spots, and the diagnosis of the alteration could imply the analysis of many areas of a gene until one alteration is found.

Moreover, the genetic alterations responsible for a specific genetic disease can vary within populations from one geographic area to another. One mutation can be responsible for almost all cases of a disease in one area or ethnic group but this may not be the case in another area. That implies that the techniques for the diagnosis of a particular genetic alteration could vary from one geographic area to another, according to the different mutations.

It is also important to consider that there could be different techniques to reliably and accurately diagnose a specific genetic alteration. The selection of the technique depends in many cases on the personal preference of the professional or on reasons such as resources available. The efficiency and costs of the resources involved are elements that have to be taken into account when deciding which diagnostic technique is more adequate for a given lab.

Labs use specific reagents for the PCR (polymerase chain reaction) and other such techniques. These reagents can be provided by a commercial company or developed by the same centre performing the test, the so-called home-brew tests. These home-brew tests (with in-house prepared materials) fall outside any regulatory frame unlike commercial in-vitro diagnostic products regulated by the IVD (in vitro diagnostics) European directive.

In Table 2, different techniques for molecular genetic diagnosis of some hereditary diseases are summarised. A much more detailed information for each disease can be found in the guidelines included in the web page of the European Molecular Genetics Quality Network (http://www.emqn.org/guidelines.htm).

Table 2. Techniques for molecular genetic diagnosis of some hereditary diseases

Disease	Techniques
Cystic fibrosis	PCR analysis Mutation analysis Direct sequencing
Duchenne muscular dystrophy	Multiplex PCR Microsatellite analysis Mutation analysis (MAPH, DHPLC, DGGE, protein truncation test [PTT], myoD-induced myogenesis)
Familial Breast cancer	PCR analysis Mutation analysis
Familial adenomatous polyposis coli	Linked-marker testing Mutation analysis (SSCP, DGGE, heteroduplex analysis, RNAse protection and chemical mismatch cleavage)
Fragile-X syndrome	PCR analysis Southern blot analysis Mutation analysis (SSCP, DGGE, CSGE) Direct sequencing
Friedreich ataxia	PCR analysis Southern blot analysis Mutation analysis Linked-marker testing
Haemophilia A	PCR analysis Southern blot analysis Mutation analysis Factor VIII gene inversion Von Willebrand Factor - Factor VIII binding analysis Linked-marker testing Direct sequencing
Haemophilia B	Linked-marker testing Mutation analysis Factor IX deletions Direct sequencing
Hereditary motor and sensory neuropathies	PCR methods based on STR markers RFLP Southern blot methods Linked-marker testing
Hereditary non-polyposis coli	RT-PCR MSI (MicroSatellite Instability) analysis Mutation analysis Direct sequencing Linked-marker testing
Huntington disease	PCR analysis Southern analysis
Myotonic dystrophy	PCR analysis Southern analysis
Prader Willi / Angelman syndromes	FISH Methylation analysis Microsatellite analysis
Spinal muscular atrophy	PCR analysis Southern analysis Mutation analysis
Spinocerebellar ataxias (other than Friedreich ataxia)	PCR analysis Southern analysis
Y-chromosome microdeletions	PCR multiplex (at least duplex)

2. Survey on genetic testing in Spain

In the context of a prospective study analysing the technical needs and options for harmonisation and quality assurance of genetic testing services in the EU, a pilot survey was performed, at the beginning of 2002, on the current situation and practices of genetic testing services for hereditary diseases in Spain. The survey aimed to assess the situation in Spain, as a case example for EU general situation. The methodology and results of the survey are presented below.

Some related topics not covered in the survey of centres have been explored by means of literature review and expert consultation and are covered in Chapters 3 and 4. These topics include the existence of specific regulations or guidelines on genetic testing in Spain, the referral pathways for patients and the question of who is responsible to provide relevant information to patients.

2.1 Objectives

- To identify centres performing genetic testing for hereditary diseases in Spain.
- To list hereditary diseases for which genetic testing is performed in Spain and the number of tests performed for each disease during the year 2001.
- To describe some laboratory characteristics: ownership and type of centre activity (health care, research or teaching).
- To detect centres of reference at the regional or state level, particularly for rare diseases.
- To explore processes of accreditation and quality assurance in those centres.
- To describe centre policies to ensure informed consent and privacy of the results.

2.2 Methodology

The following steps have been carried out:

1.- Internet search for resources on this field and Spanish associations web sites.

2.- Exploratory initial personal interviews on-site with key stakeholders, representatives of professional organisations or national experts in the area. Also telephone interviews with other key stakeholders of groups or organisations. The organisations involved in the area that were contacted are listed in Annex 1.

3.- Identification of different centres or units that perform genetic testing for hereditary diseases in Spain. To achieve this we have combined different strategies:

- Internet search in relevant on-line directories and web pages (EDDNAL European Directory of DNA Diagnostic Laboratories, Spanish Society of Human Genetics and Spanish Society for Neurology).

- General internet search using the terms "diagnóstico genético" (genetic testing).

- A request to the experts identified to check the list of centres and complete it.

- Telephone interviews with professionals performing genetic testing in all the centres identified in the previous steps, asking for other units in their centres that perform genetic testing. All persons contacted who were performing genetic testing for hereditary diseases agreed to provide their e-mail address and to fill in the questionnaire.

- A provisional list of centres previously identified was sent to all those receiving the questionnaire, and they were also asked to check for any missing centres or units, with special reference to their autonomous community.

4.- Questionnaire

A questionnaire was designed to cover all the areas included in the objectives of the study.

In order to improve the response rate, and due to the exploratory approach of the study, we decided to make a questionnaire that could be answered in ten to fifteen minutes.

The questionnaire included closed questions (yes/no type) and open-ended ones to give the opportunity to participants to express freely what they believed to be relevant.

To facilitate respondents to answer the questionnaire, different lists of conditions that could be tested by means of genetic tests were provided. Those lists were the ones used by the Spanish Society of Human Genetics (Asociación Española de Genética Humana) in the directory of centres and genetic tests included in the web page of that organisation.

An initial version was drafted and sent to some key stakeholders. A validation of the questionnaire or a pilot study has not been carried out, due to the time constraints of this study. The questionnaire is attached in Annex 2.

5.- Questionnaire was presented by telephonic contact and sent by e-mail

As explained above, professionals of the different centres or units identified in previous steps were contacted and the origin and objectives of the study were explained to them by phone.

The questionnaire was sent with a introductory letter (Annex 3) by e-mail explaining the objectives and institutions involved in this study to all the 53 centres identified. In many of them, there were several units involved in genetic testing and the questionnaire was sent to all of them, and to some other professionals identified in the previous steps.

The President of the Spanish Association for Human Genetics, Dr Isabel Tejada, and the Coordinator of the Human Genetic Section of the Spanish Society of Genetics, Dr Joan Fibla, have addressed all members of both professional associations, expressing the associations' support to this study. They also have sent them a copy of the questionnaire.

An e-mail with the questionnaire was also sent to all the members of the Working Group on Molecular Genetics in Paediatric Endocrinology, of the Spanish Society of Paediatric Endocrinology.

To improve the survey response rate, a reminder was sent three weeks after the questionnaire. Centres were contacted again, usually by telephone or by the e-mail in the cases in which telephonic contact with the responsible person was not possible.

2.3 Results of the survey

2.3.1 Characteristics and geographic distribution of the centers identified

53 centres performing genetic testing for hereditary diseases have been identified. The term "centre", as used in this context, means institutions (i.e. hospitals or universities) as a whole. Those hospitals or universities with more than one unit performing genetic testing have been considered as a single centre for the purpose of this study.

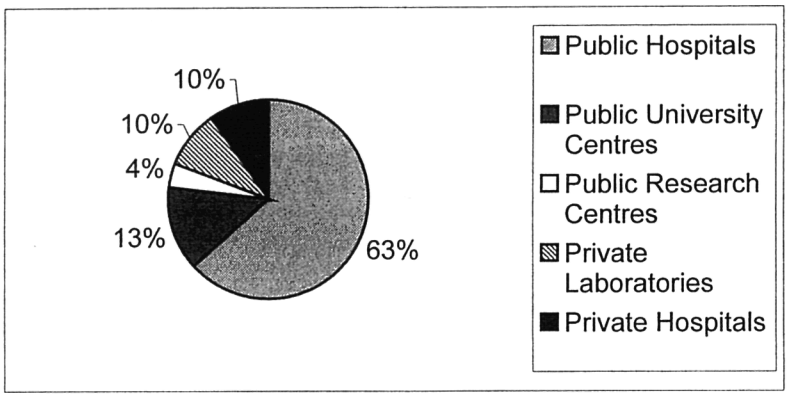
The list of centres identified, units involved and contact persons, their postal and e-mail addresses and the telephone number, can be found in Annex 4.

All 53 centres (100 %) answered the questionnaire. Taking into account the voluntary participation of the centres, this response rate probably reflects both the collaborative attitude of those involved in genetic testing and the need for an updated directory of centres doing those tests, as it has been expressed by many professionals contacted. Table 3 and Figure 1 show the distribution by type of centre.

Table 3. Characteristics of Spanish centres doing genetic testing

	Public	Private	Total
Hospitals	33	5	38
Universities	7	-	7
Research centres	2	-	2
Laboratories	-	6	6
	42	11	53

Figure 1. Types of centres performing genetic tests in Spain, by percentages



The number of centres identified in each autonomous regions is shown in Table 4, including population size and the number of conditions tested in each region. There is at least one centre performing genetic testing in each autonomous region in Spain.

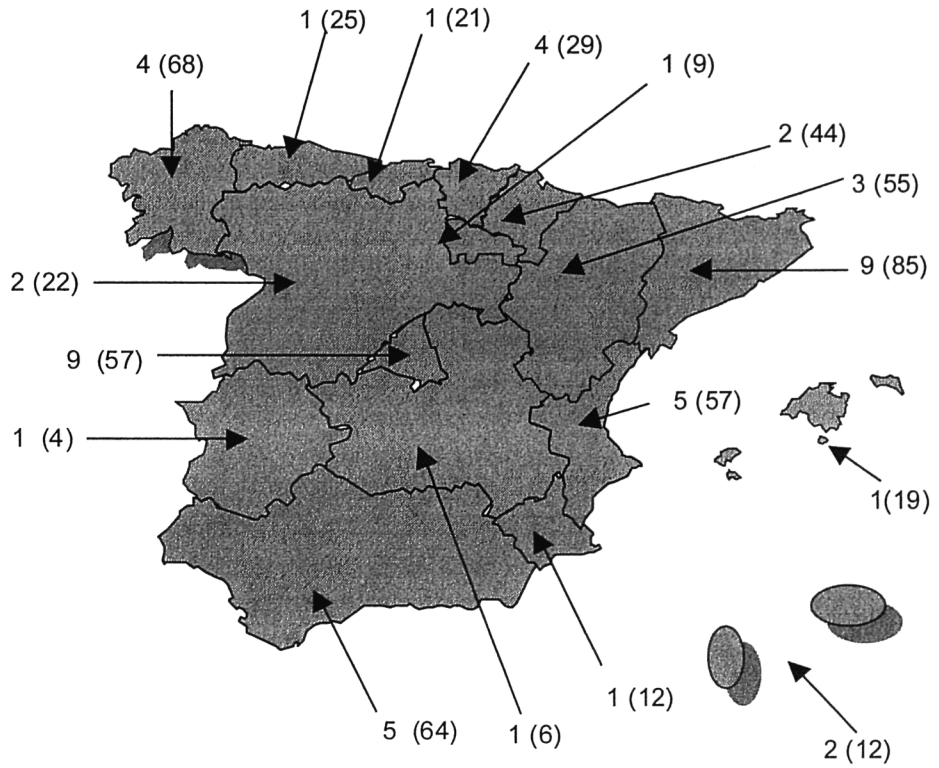
Table 4. Distribution of centres and conditions tested by Regions

REGIONS	N. centres	Population in 2001*	Conditions tested
Andalusia	6	7,403,968	64
Aragon	3	1,199,753	55
Asturias	1	1,075,329	25
Baleares	1	878,627	19
Canarias	2	1,781,366	21
Cantabria	1	537,606	21
Castilla-La Mancha	1	1,755,053	6
Castilla Leon	2	2,479,425	22
Catalonia	9	6,361,365	85
Comunidad Valenciana	5	4,202,608	57
Extremadura	1	1,073,381	4
Galicia	4	2,732,926	69
Madrid	9	5,372,433	57
Murcia	1	1190,378	12
Navarra	2	556,263	44
Basque Country	4	2,101,478	29
Rioja	1	270,400	9
Total Spain*	53	41,116,842	214

* Source: I.N.E. Total includes two autonomous cities Ceuta (75,694) and Melilla (68,789)

The Spanish health system has a high level of decentralisation. There are 17 Autonomous Regions with Regional Health Services, fully responsible for services provision and financing. As it can be seen in Table 4, the geographical distribution of the number of centres doing genetic testing does not directly correlate with the population size of the regions.

Figure 2. Distribution of centres and genetic conditions (in brackets) tested by centres in each Spanish region



Catalonia and Madrid have a large proportion of centres, with nine centres in each region. Their population is smaller than that of Andalusia, where only 5 centres are doing genetic tests. In part, this can be explained by the presence of some private centres in Catalonia and some national research centres located in Madrid.

In regions such as Aragon, with a population of 1,199,753 persons, 3 centres are doing genetic tests for 53 conditions. In Galicia, with a population of 2,732,926 persons, 4 centres are doing tests for 68 conditions. Basque Country is also a region with small population and in comparison a high number of type of tests offered.

These imbalances in the distribution of centres and conditions tested probably reflects the unplanned development of genetic testing in Spain and the existence of some units very active in research in some autonomous regions.

The consequences of this uneven distribution of centres and conditions tested between regions on possible differences in the accessibility to genetic testing have not been analysed. This topic would deserve more in-depth research.

2.3.2 Genetic conditions tested

According to the data obatained in the questionnaires, 214 genetic conditions are currently being tested in Spain while EDDNAL, the European directory contains 580 genetic conditions tested in European centres.

Detailed results for all the conditions are shown in Annex 5, classified by functional areas (neurological diseases, haematological diseases, metabolic problems, hereditary cancers, collagen diseases, primary immune-deficiencies, mitochondrial diseases and others).

In Annex 6, results are presented grouped by autonomous regions. The number of tests performed for each disease, in each centre, in the year 2001 is shown where available. For some centres the precise number of tests for a specific condition is missing, although it is known that the test is available at the centre.

Figures of tests for each condition include those performed for research purposes and those for genetic counselling in the context of clinical practice. These data are estimated from survey responses, and therefore should be interpreted with caution.

It is also possible that data from some specific units within centres are missing. It could be due to either lack of information about specific subunits in the identification phase or non-response to the questionnaire.

In Table 5, summary data on the 15 conditions most frequently tested in 2001 are presented. They correlate with those with a higher frequency in the population (hemochromatosis, coagulation disorders, fragile X, cystic fibrosis or haematologic cancers), also having a higher interest and validity for clinical purposes. Also, it is not surprising that testing for those frequent conditions is done in numerous centres and regions. The mutations responsible for the diseases are well known and the techniques are reliable and relatively easy to learn.

Research trends, however, also influence which conditions are being tested more often, as a reflection of the close relationship between research and clinical application in the field. Centres investigating a particular disease will carry a higher number of tests for it, reflecting a higher sample size for a particular test even though the alteration is not more common nor more often requested by physicians. Hence, genetic testing for Alzheimer disease, for example, was one of the most frequently performed tests in the year 2001, though it currently has little clinical usefulness. According to experts, it is an area of very active research and many tests are performed.

Table 5. Conditions most frequently tested in Spain in the year 2001

	N. tests	Centres	Regions
Hemochromatosis	4398	26	13
Thrombophilia (Mut. G1691A of Leiden's factor V gene) (Mut G20210A of prothrombin gene)	3644	15	11
Prothrombin gene	2756	14	9
Factor V Deficiency	2597	16	10
Fragile X Syndrome	2613	24	14
Cystic fibrosis	1900	28	14
Leukemia and lymphoma	1881	11	7
Homocystinuria	1355	6	4
Myotonic dystrophy Steinert	1055	22	12
Apolipoprotein B (APOB) deficiency	932	3	3
P 53 exons 4,5,6,7,8	888	13	9
21-hydroxylase deficiency	866	6	4
Alzheimer (Presenilin 1, APP, APOE)	842	15	11
Azoospermia and oligospermia	766	18	11
Dominant Ataxias (ADCAS) SCA1; SCA2 (Machado), SCA3; SCA6; SCA7; SCA8; SCA9; SCA10; SCA12	630	12	8

2.3.3 Reference centres

A question was included in the survey about reference centres at regional or national level (data presented in Table 6). Due to the exploratory nature of the survey and for the sake of brevity, questions on specific diseases for referral were not included in the questionnaire.

At least 36 centres considered themselves as reference centres at the regional or national level. This probably shows that when a centre starts doing testing for one disease it usually ends up being the only one doing it at the regional level and in the case of less frequent conditions becomes reference centre at a national level.

Table 6. List of centres of reference

Centre	Regional or national reference centres	Diseases
Hospital Virgen del Rocío Sevilla	Not stated	Not stated
H. San Cecilio Granada	Not stated	Not stated
Hospital Carlos Haya Málaga	Not stated	Acute promielocitic leukaemia, acute lymphoblastic leukaemia Philadelphia positive
Hospital Miguel Servet Zaragoza	Not stated	Not stated
Universidad de Zaragoza	Not stated	Not stated
Centro de Analisis Genéticos Zaragoza	Not stated	Not stated
Hospital Central de Asturias	Regional and national	Not stated
Hospital Son Dureta. Mallorca	Not stated	Not stated
Hospital Valdecilla Santander	Not stated	Not stated
Universidad de La Laguna Tenerife	Regional and national	Primary hyperoxaluria
Hospital Materno Infantil de Badajoz	Not stated	Not stated
Centro de Patología Celular Barcelona	Not stated	Not stated
Hospital Clínico de Barcelona	Regional and national	Not stated
Hospital San Juan de Dios Barcelona	National	Rett, CMT1A, HNPP
Centro Genética Médica y Molecular –IRO	Regional and national	Not stated
Hospital del Mar - Facultad de Ciencias de la Salud (Universitat Pompeu Fabra)	National	Leukaemia and Lymphoma (Hosp del Mar) Williams Syndrome (UPF).
Hospital Parc Taulí	Regional and national	Not stated
Hospital Valle Hebron	Regional and national	Male pseudo hermaphroditism (mutations in genes AR, SRD5A2 y 17HSD3)
Hospital San Pablo Barcelona	Regional and national	Not stated
Centro Oncológico de Galicia	Regional	Not stated
Hospital de Conxo	Regional	Not stated
Hospital Juan Canalejo La Coruña	Regional, national and international	Von Willebrand
Hospital Clinico de Santiago	Regional	Not stated
Hospital La Paz Madrid	Regional, national and international	Cystis fibrosis, miotonic dystrophy, alfa1antitripsina genotyping, Duchenne muscle dystrophy, Huntington disease, hemochromatosis, 21-hidroxilasa deficiency, achondroplasia, GH receptor deficiency, acute intermittent porphyry, erithropoietic congenital porphyry, MODY type diabetes.
H. Ramón y Cajal Madrid	National	Not stated
Centro de Diagnóstico de Enfermedades Moleculares - U. A. de Madrid	Not stated	Not stated
Fundación Jiménez Díaz Madrid	Not stated	Not stated
Hospital Gregorio Marañón Madrid	Regional and national	21-hidroxilasa deficiency, 11-hidroxilasa deficiency, Turner syndrome, discondrosteosis, achondroplasia, small height with skeleton disproportion, hypochondroplasia
Hospital 12 Octubre Madrid	Not stated	Not stated
Centro de Investigación sobre Anomalías Congénitas Madrid	National	Not stated
Centro Nacional Investigaciones Oncológicas (CNIO) Madrid	Not stated	Not stated
Clinica Universitaria Pamplona	National	Not stated
Hospital de Basurto	Regional and national	Fragile X
Hospital de Cruces Baracaldo	Regional and national	Not stated
Hospital Donostia	Not stated	Not stated
Hospital La Fe. Valencia	Not stated	Not stated

2.3.4 Accreditation related topics

Accreditation systems could target the professionals and/or the centres performing the tests. Regarding professionals, there is no biomedical specialty or university degree on human genetics in Spain. This is one of the problems mentioned by many in the interviews and questionnaires.

The Spanish Society for Human Genetics has a procedure to accredit individual professionals (http://www.uam.es/otros/AEGH/paginas/acreditacion.html). These professionals must be members of the association, for at least two years, and either a medical doctor, a biologist, a pharmacist, a chemist or a biochemist. They must develop their professional activity in the area of human genetics. A group of experts appointed by the society analyses the curriculum of the applicants and by means of an explicit list of areas considered, scores the different merits of the applicant, providing the accreditation if he/she reaches a certain level. This process is voluntary and has no legal backing or support from official institutions. To date, about 180 professionals have passed the procedure.

Regarding the centres or diagnostic laboratories, in Spain there is no specific system for the accreditation or control of the centres which perform any kind of genetic test. In some autonomous regions, laboratories dealing with clinical analysis are subject to an accreditation process for biological testing, but it is not an iterative process with periodic re-evaluation. 17 Spanish centres out of the total sample surveyed are voluntarily included in the EDDNAL directory and almost all of them are active participants.

2.3.5 Quality assurance related topics

Some of the centres are carrying out quality assurance activities for specific tests or diseases. Of those centres registered in EDDNAL, some are participating in quality control schemes developed by the EMQN or the Cystic Fibrosis European Thematic Network. Those who stated so in the questionnaire are listed in Table 7.

At least two centres are in the process of getting the ISO 9000 – 9002 qualifications (Hospital Materno Infantil Las Palmas, Hospital Clínico Barcelona).

2.3.6 Informed Consent and Privacy of Data

According to survey replies, informed consent of the patient is requested by 90% of centres. Usually, the physician requesting the test is the one who normally obtains it from the patient. In tests performed in the context of research studies the researchers are responsible for the informed consent process.

According to the survey, almost all centres have developed specific policies to warrant the privacy of the tests results.

Table 7. List of centres of reference participating in Quality Assurance schemes

Disease	Centres
Ataxias	Centro Genética Médica y Molecular -IRO
Breast cancer	Hospital San Pablo Barcelona
Cystic Fibrosis	Centro de Análisis Genéticos Zaragoza Centro de Bioquímica y Genética Clínica de Murcia Centro de Patología Celular Barcelona Hospital La Fé Valencia Hospital La Paz Madrid Hospital Materno-Infantil Las Palmas Hospital San Juan de Dios Barcelona Hospital San Pablo Barcelona Hospital Son Dureta Mallorca Institut Reserca Oncológica Instituto Biología Genética Molecular. Valladolid Oncológico de Galicia
Duchenne/Becker	Hospital San Pablo Barcelona
Fragile X syndrome	Centro de Bioquímica y Genética Clínica de Murcia
Huntington	Hospital de Conxo Santiago
Prader Willi – Angleman	Centro de Bioquímica y Genética Clínica de Murcia

2.3.7 Financing of the tests and related topics

According to survey replies, more than 95% of the test are financed by public institutions, whether hospitals or universities.

Experts mentioned that there is no earmarked money for these tests, except when tests are performed in the context of specific research studies, and in these cases the financing of the tests is usually limited to the duration of the study.

The average cost of performing one test is in most cases very dependent on the number of tests performed in one centre for a specific disease. Average costs of genetic tests could be around €240, ranging from €120 for cystic fibrosis to €330 for a study for ataxia, according to the estimate of one expert.

3. Genetic testing products on the market

In order to get information on commercial tests available in Spain, different sources were contacted and surveyed :

- Ministry of Health, Department of Pharmacy and Health Products. No list of products is currently available, but there is a Royal Decree on health products for in vitro testing (R.D.1662/2000) due to enter into force before the end of 2003. This is further explained in section 4.2. This Decree will make compulsory for all new products to be sold in Spain to have CE mark and to be registered, but no further information was available when carrying the survey.

- FENIN, the Spanish Federation of Health Technology Industries (Federación Española de Empresas de Tecnología Sanitaria: http://www.fenin.org) is represented in the European Diagnostic Manufacturers Association, EDMA (http://www.edma-ivd.be/), Executive Committee. It comprises 40 companies involved in *In Vitro* Testing, but there is neither a specific directory of companies marketing products, nor a list or database with commercial products for genetic testing of hereditary diseases in Spain.

- Consultation with professionals involved in the field. Information on most frequent products and companies was provided and these were contacted.

From these sources, it was only possible to get incomplete information. Most of the centres and labs performing genetic testing only test for a small number of conditions, and in many cases tests are home-brew. Various commercial companies are marketing reagents or kits specially designed to help professionals in their job, mainly covering those diseases with higher incidence.

From these different information sources consulted, 16 companies marketing various products related with genetic testing in Spain have been identified. Nine of them are European companies:

- Amersham (UK)
- Eppendorf (Germany)
- Innogenetics (Belgium)
- MWGAG (Germany)
- Novagen (Germany)
- Oncor (France)
- Orchid Biosciences (UK)
- Qiagen (Netherlands-Germany)
- Roche (Switzerland)]

and seven USA based companies:

- Applied Biosystems
- Axygen, Beckman Coulter
- Bio Rad

- Genpak Vysis
- Licor Biosciences
- Promega
- RNWAY

Table 8 shows the diseases for which specific reagents or kits are marketed in Europe.

Table 8. Commercially available In Vitro Tests for the detection of gene and chromosome alterations*

Inborn Gene or Chromosome Alterations	
Monogenic Disorders	**Polygenic Disorders**
Cystic Fibrosis	Alzheimer's Disease
Duchenne Muscular Dystrophy	Asthma
Factor V Leiden	Atherosclerosis
Fragile X Syndrome	Diabetes
Hemochromatosis	Hypertension
Haemophilia	Multiple Sclerosis
Huntington Chorea	Osteoporosis
Polycystic Kidney Disease	(Other)
Sickle Cell Anemia	
Tay Sachs Disease	
Thalassaemia	
Friedreich's Ataxia	
Spinocerebellar Ataxias Type 1,2,3,6,7,8	
Prothrombin Mutation	
(Other)	

Chromosomal Disorders	**Acquired Gene or Chromosome Alterations**
Down's Syndrome	p53
Edwards Syndrome	k-ras
Klinefelter syndrome	BRCA 1+2
Pätau Syndrome	Ret
Turner Syndrome	c-myc
Other Chromosomal Disorders Tests	HNPCC
	APC
Polymorphisms	t(9;22)
HLA-Typing	t(8;14)
(Other)	t(14;18)
	inv (16)
	Telomerase
	HER-2/neu
	Others

* (Source: http://www.edma-ivd.be/Classification/Final_version_5.xls, access 2002-03-22)

Table 9 shows a list of diseases for which commercial products are marketed in Spain and the companies producing them. This list has been elaborated from catalogues, commercial leaflets, internet web pages and telephonic contacts with company representatives.

Table 9. Companies marketing kits or reagents for genetic conditions

Monogenic Disorders	**Companies marketing kits or reagents**
Alpha Thalassaemia	BIO-RAD
Alpha 1 antitrypsin	ORCHID BIOSCIENCES
Beta Thalassaemia	BIO-RAD
Cystic Fibrosis	APPLIED BIOSYSTEMS; INNOGENETICS; SAVYON
Factor V Leiden	BIO-RAD; ORCHID BIOSCIENCES
Hemochromatosis	BIO-RAD; SAVYON
Leukemia	BIO-RAD; VYSIS
Lymphoma	VYSIS
Prothrombin mutation	ORCHID BIOSCIENCES
Sickle Cell Anemia	BIO-RAD
Tay Sachs disease	ORCHID BIOSCIENCES
Polymorphisms	
HLA-Typing	INNOGENETICS; APPLIED BIOSYSTEMS; ORCHID BIOSCIENCES
Apolipoprotein E	INNOGENETICS
Acquired Gene or Chromosome Alterations	
HER-2/neu	VYSIS
Associated Disease (Gene)	
Predisposition to breast & ovarian cancer HER-2/neu (Brca1) (Brca2)	SAVYON –Myriad
Hypercholesterolemia, cardiovascular disease (Apolipoprotein E)	SAVYON –Myriad
Predisposition to colorectal cancer	SAVYON –Myriad
Predisposition to thrombosis	SAVYON –Myriad
Differential Diagnosis	
Familial Mediterranean Fever	SAVYON –Myriad

4. Other topics

Some topics not covered in the questionnaire have been explored through interviews with experts and are presented below.

4.1. Access for patients to genetic testing and counselling

The referral of a patient to genetic testing in Spain does not follow agreed protocols. The patient might have access to genetic testing through the regular clinical channel (within the health-care system) or as part of a research activity.

On the one hand, in the clinical set-up, genetic testing follows referral by a physician. Usually, the referral is done by a medical specialists in a hospital requiring confirmation of the diagnosis of a genetic disease after clinical examination or biochemical testing.

The referral to the laboratory is not necessarily accompanied by a request for genetic counselling by a specialist in genetics, and the physician is him/herself responsible for providing the relevant information to the patient and his/her relatives. That depends on the existence of specific genetic units responsible for counselling of patients and his/her relatives and whether specific agreements between clinicians and genetic units are in place.

A common concern is that a positive result of the genetic test, the confirmation of the genetic condition, is not always followed by the offer of genetic counselling to the patient and relatives.

In this clinical set-up, the physician who orders the tests is also the person responsible for requiring informed consent from the patient. The labs may not have this consent available when performing the genetic test.

On the other hand, many times, patients have access to genetic testing as part of research activities on inherited diseases. The researchers might look for patients in a pro-active way. The research team, usually including physicians and members of genetic or research units, has a research protocol that has been approved by either the funding institution or a specific committee in the hospital. The research protocol must define the scope of the study and procedures related to the information to be provided to patients post-testing. The researchers are also responsible for requiring informed consent from participants in the study.

4.2 Regulatory developments in genetic testing in Spain

The information of this section has been obtained from a consultation with Ministry of Health officers, key documents on the regulation of health technologies in Spain and interviews with experts in the field.

Regulation and authorisation of *in-vitro* diagnostic medical devices are under the jurisdiction of the Deputy Directorate of Medical Devices, General Directorate of Pharmacy and Medical Devices, Ministry of Health and Consumers Affairs. Directive 98/79/EC, on *in-vitro* diagnostic medical devices, has been transposed into national legislation by the Spanish government in the form of a Royal Decree 1662/2000, approved on 29 September 2000 by the Spanish Health Ministry. The provisions of this Decree will enter into force before the end of 2003.

Currently, pre-market authorisation is not needed for most *in-vitro* diagnostic medical devices. An exception is testing for the detection of infection markers of a human virus of the Retroviridae family, including antigens and antibodies related to AIDS.

Apart from that, no other specific legal regulations exist on genetic testing in Spain, neither at state level nor at regional level. A specific committee on genetic testing does not exist. Some aspects of relevance to genetic testing have been considered by other committees in the Ministry of Health, such as those involved in ethics, clinical trials, in-vitro fertilisation or oncology.

According to these data and survey results, the introduction of genetic testing in a centre does not require previous official authorisation in Spain, the decision being taken by

each hospital. The appropriate selection of tests is part of the analyst's professional responsibility. This could explain the multiplicity of centres and diversity of tests in different regions, and the lack of formal coordination platforms between centres or a centralised register.

4.3 Guidelines

We have not found specific guidelines developed in Spain in relation to genetic testing for specific conditions. Several university departments, hospitals and other research bodies are working on aspects of the topic, but these initiatives are not centralised or co-ordinated in a systematic way.

The Medical Association of Barcelona (Colegio Oficial de Médicos de Barcelona) has recently produced a document with recommendations for genetic testing. They stress the importance of informing the patients of the clinical validity of the genetic test before doing it. The recommendations cover relevant topics such as informed consent, privacy of the results, genetic counselling, participation in screening programs and ethical considerations[3].

4.4 Rare diseases

An specific area of concern is the accessibility to genetic testing for rare diseases. Once the clinical diagnosis points towards a given disease, the specialists usually contact the genetic unit in their region or some centre of reference at state level that indicates where to refer the patient for testing. According to experts opinion, an important difficulty is to actually reach the clinical conclusion that a patient or a family has a rare disease in which a genetic disorder plays an important role.

Payment for the tests done in other centres, either in Spain or abroad, could be in some cases cumbersome due to the bureaucratic mechanisms, but nevertheless the tests are are, in their majority, carried out, as stated by experts.

There seem to be unofficial agreements between professionals of different centres not to charge for testing for rare diseases for which only one or two cases a year are referred for testing. In those cases the workload involved is not important and the professionals of the lab usually are engaged in research networks interested in examining a number of patients with those rare diseases. They need cases for their studies and to have the experience necessary to reach and maintain expertise and proficiency in testing for those diseases.

In more prevalent diseases, the workload for testing will be really relevant and labs either try to charge for the testing or deny access to patients referred from other centres.

[3] http://www.diariomedico.com/edicion/noticia/0,2458,126081,00.html, last access 26 March 2002

Acknowledgements

The authors wish to thank Dr. Isabel Tejada (President of the Spanish Society for Human Genetics) and Dr. Federico Volpini (Spanish representative in the European Molecular Genetics Quality Network) for their support and for the external review of this report.

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Annex 1. Organitations involved in the area

Asociación Española de Genética Humana, member of the European Society of Human Genetics).
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Sociedad Española de Errores Innatos del Metabolismo
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Registro Español de Malformaciones Congénitas
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Federación Española de Enfermedades Raras (FEDER). Member of the European Organisation for Rare Disease (http://www.eurordis.org)

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President: Moisés Abascal Alonso
PARD I Coordinator: Rosa Sanchez de Vega

Sociedad Española Neurología http://www.sen.es/recursos/genet/genetica.html

Annex 2. Questionnaire sent to the centres and laboratories

Encuesta sobre la oferta de diagnóstico genético para enfermedades hereditarias en España

La presente encuesta es parte del estudio "Prospective study on Genetic Testing Services quality assurance and harmonisation in EU" del Institute for Prospective Technological Studies, Joint Research Centre de la Comisión Europea. Esta encuesta está siendo realizada por la Agencia de Evaluación de Tecnologías Sanitarias de Andalucía por encargo de esa institución.

El objetivo de esta encuesta es obtener información actualizada sobre los centros que realizan algún tipo de prueba de diagnóstico genético para enfermedades hereditarias en España.

Le rogamos rellene directamente los datos en este mismo archivo y nos lo envíe por E-mail.

Centro: Unit: Contact person: Address: Telephone: E-mail: Type of centre: (sanitario, universitario, ... ; de propiedad pública, privada, concertado,...)

1.- ¿Realiza su centro algún tipo de prueba de diagnóstico genético de enfermedades hereditarias? (SI ó NO)

En caso de no realizar en la actualidad ese tipo de pruebas pero sí en el pasado no tiene que contestar el resto de preguntas de esta encuesta. Simplemente díganos que pruebas realizaban y cuando dejaron de hacerlas.

2.- De las siguientes listas con diversas enfermedades hereditarias, señale aquellas para cuales de ellas disponen o realizan pruebas genéticas en su centro y el número de pruebas que ha realizado el pasado año para cada enfermedad.

En caso de realizar alguna prueba que no aparezca en las tablas siguientes escríbalo a continuación o en la última fila de cada tabla.

PATOLOGÍAS NEUROLÓGICAS	SI/NO (N° de pruebas en 2001)
Ataxias dominantes (ADCAS) SCA1; SCA2 (E. de Machado); SCA3; SCA6; SCA7; SCA8; SCA9; SCA10; SCA12	
Ataxia Friedrich (X25)	
Atrofia dentato-rubal palidoluisiana (Dentatorubro-palidoluisiana)	
Atrofia muscular espinal (Werdnig-Hoffmann y Kugelberg-Welander) (SMN)	
Atrofia muscular espino-bulbar Enfermedad de Kennedy. (AR, exón 1)	
Demencia frontotemporal (tau) (detección de mutacions) / Parálisis supranuclear progresiva tau (genotipo polimórfico)	
Distrofia facio-escapulo-humeral	
Distrofias maculares dominantes (gen DS/periferina)	
Distrofia miotónica de Steinert	
Distonía de Torsión (DYT1)	
Distrofia muscular de Duchenne/Becker	
E. Hirschsprung	
E. Alzheimer (Presenilina 1, APP, APOE)	
E. Huntington	
E. Norrie	
Neurofibromatosis tipo 1	
Neuropatía Charcot-Marie-Tooth	
Análisis de ligamiento de las NF1 y NF2	
Neuropatía óptica de Leber	
Neuropatía por sensibilidad a la presión	
Paraplejla espástica familiar	
Parkinson (Parkin, a-sinucleina)	
Retraso mental asociado a la fragilidad tipo FRAXE (FMR2)	
Retraso Mental (GDI1)	
Retraso Mental (PAK3)	
S°. Angelman	
S°. Prader Willi	
S°. Smith-Magenis	
S°. Rett	
S°. de Williams-Beuren (LOH , FISH) (ATP7B)	
S X-Frágil (FMR1)	
OTRA(S) ESPECIFICAR	

PATOLOGÍAS HEMATOLÓGICAS	SI/NO (Nº de pruebas el año 2001)
Hemofilia A	
Hemofilia B	
E. Wisckott-Aldrich	
Leucemias	
a-Talasemia	
ß-Talasemia	
Trombofilia Hereditaria (Mut. G1691A del gen del factor V Leiden) (Mut G20210A del gen de la protrombina	
OTRA(S) ESPECIFICAR	

CÁNCER HEREDITARIO	SI/NO (Nº de pruebas el año 2001)
Cáncer mama (BCRA1 y BCRA2)	
CK-19 (Células circulantes, cel epitelial, mama, ovario)	
GST mu (colon, vejiga)	
K-RAS (CODON 12, 13)(pulmón, colon, ovario, laringe)	
PSA (cel. Circulantes sangre, ca. próstata)	
P53 EXONES 4,5,6,7,8 (Pulmón, Mama, Ovario, Endomet.)	
E. Li-Fraumeni (p53)	
Feocromocitoma familiar	
Melanoma familiar (pl5, p19,CDK4)	
Melanoma familiar (pl6)	
MEN1	
Neoplasia endocrino múltiple MEN2A	
Neoplasia endocrino múltiple MEN2B	
Poliposis adenomatosa familiar	
E. Von Hippel Lindau	
OTRA(S) ESPECIFICAR	

PATOLOGÍAS METABÓLICAS	SI/NO (Nº de pruebas el año 2001)
Acidemia glutárica tipo I	
Acidemia propiónica	
Adrenoleucodistrofia ligada al X	
Déficit de Acil-CoA deshidrogenasa de cadena media (MCAD) (Mutación K304E)	
Defecto familiar de ApoB	
Déficit de 21-hidroxilasa	
Déficit de 11-beta-hidroxilasa	
Déficit de 17-alfa-hidroxilasa	
Déficit de 3-beta-hidroxiesteroide-deshidrogenasa	
Déficit de alfa-1-antitripsina	
Def. Carnitina Palmitoil Transferasa II (Mutación Y628S)	
Déficit de GH	
Déficit de hidroxiacil-CoA deshidrogenasa de Cadena larga (LCHAD)	
Diabetes tipo MODY. (GK, HNF1)	
E. Gaucher	
E. de Hurler	
E. de Morquio B	
E. Sanfilippo A	
E. Sanfilippo B	
E. Tay-Sachs	
Galactosemia	
Fenilcetonuria-Fenialaninemia (PAH)	
Hiperaldosteronismo primario tipo I	
Hiperplasia suprarrenal congénita	
Leucodistrofia Metacromática y pseudodeficiència Arilsulfatasa A	
Ligamiento a LDL (Hipercolesterolemia)	
Sº de insensibilidada la GH	
OTRA(S) ESPECIFICAR	

ENFERMEDADES DEL COLÁGENO	SI/NO (Nº de pruebas el año 2001)
Ehier-Danios	
Osteogénesis imperfecta	
OTRA(S) ESPECIFICAR	

OTRAS PATOLOGÍAS	SI/NO (Nº de pruebas el año 2001)
Azoospermia y oligospermia (Microdeleciones cr. Y)	
Acondroplasia/Hipocondroplasia (FGFR3)	
Coroideremia	
Fibrosis Quística (CFTR)	
Cistinuria	
Déficit de Hormona de Crecimiento. (GH)	
Enfermedad celiaca	
Enfermedad de Norrie (NDP)	
Enfermedad de Wilson (ATP7B)	
Factor V de Leyden	
Gen de protrombina	
Gen SRY (Alteraciones del desarrollo sexual)	
Fiebre Mediterránea Familiar	
Hemocromatosis (HFE)	
Hematuria familiar benigna	
Hipertensión familiar	
Hiper/Hipotiroidismo familiar. (receptor de TSH)	
Hipoplasia adrenal congénita	
Homocistinuria	
Holoprosencefalia	
Ligamiento a NPC1	
Lipofuscinosis Neuronal Ceroidea /Batten disease: (CLN1, CLN2, CLN3, CLN8)	
Nefronoptisis (NPHP1)	
Panhipopituitarismo	
Pseudohipoparatiroidismo	
Poliendrocrinopatia autoinmune tipo 1	
Poliquistosis renal dominante tipo 1	
Poliquistosis renal recesiva tipo 2	
Raquitismo vitamino-D-resistente	
Resistencias Androgénicas: Síndrome de Morris, Síndrome de Reifenstein (AR exones 2-8)	
Retinosis pigmentaria	
Retinosis pigmentaria ligada al X	
Retinosis pigmentaria autosómica dominante (genes RHO y RDS)	
Retinosquisis	

Síndrome de Alport (autosómico dominante, autosómico recesivo; ligado al cromosoma X)	
Sorderas esporádicas y autosómicas recesivas (Gen de la Conexina 26)	
S. Di George	
OTRA(S) ESPECIFICAR	

3.- ¿Ha pasado su centro o servicio por un proceso de acreditación específico para la realización de esas pruebas de diagnóstico genético de enfermedades hereditarias? (SI ó NO):

En caso afirmativo: ¿qué institución les ha acreditado?

4.- ¿Disponen de algún laboratorio de referencia para control de calidad de esas pruebas? (SI ó NO):

En caso afirmativo escriba el nombre de dicho laboratorio:

y señale con qué frecuencia realizan esos controles de calidad:

5.- ¿Es el suyo un centro de referencia para otros centros que le remiten muestras para pruebas genéticas de alguna enfermedad hereditaria a nivel autonómico o estatal?

6.- ¿Tienen un laboratorio u otros laboratorios de referencia para enviar muestras a analizar con tests de los que no disponen en su laboratorio? (SI ó NO):

En caso afirmativo escriba el nombre de dicho(s) laboratorio(s):

7.- ¿Hay algún caso en que sean los propios pacientes quienes financien los costes de las pruebas o son siempre financiadas directamente por la institución con cargo a fondos públicos?:

¿qué porcentaje de pruebas estima que son de financiación privada?

8.- ¿Exigen la firma de un documento de consentimiento informado antes de la realización de las pruebas? (SI ó NO):

9.- ¿Tienen una política expresa para garantizar la privacidad de los datos que analizan? (SI ó NO):

Si quiere añadir o sugerir alguna otra cosa, hágalo a continuación.

Envíenos por favor este archivo a la siguiente Address: jrueda@euskalnet.net.

MUCHAS GRACIAS POR SU COLABORACIÓN.

Annex 3. Introductory letter attached to the questionnaire

Estimad@ Dr/a:

Adjunto le remito un documento en formato Word con una encuesta sobre el uso de pruebas de diagnóstico genético de enfermedades hereditarias en España. Forma parte del estudio "Prospective study on Genetic Testing Services quality assurance and harmonisation in EU" del Institute for Prospective Technological Studies, Joint Research Centre de la Comisión Europea. Esta encuesta está siendo realizada por la Agencia de Evaluación de Tecnologías Sanitarias de Andalucía por encargo de esa institución.

Rellenarla le llevará entre 10 y 15 minutos. Contéstela directamente en el archivo de texto y enviénoslo por E-mail.

Esta encuesta va a ser enviada a todos los centros sanitarios o universitarios de los que tenemos conocimiento que realizan algún tipo de prueba de diagnóstico genético. A continuación tiene una lista de los centros que tenemos localizados. Si conoce algún otro centro, en especial de su comunidad autónoma, que no esté recogido en la lista anterior díganoslo.

Si tiene cualquier duda, no dude en ponerse en contacto con nosotros.

Dr. Jose Ramón Rueda, tf.: 944102684
Dr. Eduardo Briones, tf.: 955006841

Muchas gracias por su colaboración.

ANDALUCÍA:
Hospital Reina Sofía. Córdoba
Hospital Virgen de las Nieves. Granada
Hospital Universitario. Granada
Centro de Estudios Genéticos de Andalucía. Granada
Hospital Materno-Infantil Málaga
Hospital Universitario Virgen de la Macarena Sevilla
Hospital Universitario Virgen del Rocío Sevilla

ARAGÓN:
Hospital Clínico Universitario. Zaragoza
Hospital Miguel Servet. Zaragoza
Centro de Análisis Genéticos. Zaragoza

ASTURIAS:
Hospital Central de Asturias. Oviedo

BALEARES:
Hospital Son Dureta. Palma de Mallorca
Centro de Detección Precoz y Consejo Genético. Palma de Mallorca

CANARIAS:
Unidad de Genética Hospital Materno Infantil. Las Palmas de Gran Canaria
Facultad de Medicina. La Laguna. Tenerife

CANTABRIA:
Hospital Valdecilla. Santander

CASTILLA-LA MANCHA:
Hospital Nacional de Parapléjicos. Toledo
Hospital General de Segovia. Segovia

CASTILLA-LEON:
Instituto de Biología y Genética Molecular. Facultad de Medicina. Valladolid

CATALUÑA:
H.Materno Infantil Vall d'Hebron. Barcelona
H. la Santa Creu i Sant Pau. Barcelona
Hospital Clinic. Barcelona
Hospital San Juan de Dios. Barcelona
Instituto Bioquímica Clínica. Barcelona
Centro Patología Celular y Diagnóstico Prenatal Barcelona
Prenatal Diagnosis Barcelona
Laboratorio de Análisis Dr. Echevarne Barcelona
Instit. Resercha Oncológica. Hospitalet
Consorci Hospitalarl Parc Tauli. Sabadell
Laboratorio Cerba,S.A.E. Sabadell

C. A. VASCA:
Hospital de Basurto. Bilbao
Hospital de Cruces. Baracaldo
Policlínica Guipúzcoa San Sebastián
Instituto Neurobiología Molecular. San Sebastián

EXTREMADURA:
Hospital Materno- Infantil. Badajoz

GALICIA:
H. Materno-Infantil Teresa Herrera. La Coruña
Centro Oncológico de Galicia. La Coruña
Hospital General de Galicia & Hospital Gil Casas. Santiago de Compostela
Hospital de Conxo. Unidad de Medicina Molecular (INGO). Santiago de Compostela

MADRID:
Fundación Jiménez Díaz. Madrid
Hospital Ramón y Cajal. Madrid
Hospital La Paz. Madrid
Hospital 12 Octubre. Madrid
Hospital Clínico San Carlos. Madrid
Hospital de Móstoles.
Centro Diagnóstico Enfermedades Moleculares. Dpto. Biología Molecular. UAM.
Centro Nacional Investigaciones Oncológicas. Majadahonda

MURCIA:
Hospital Virgen de la Arrixaca. Murcia.

NAVARRA:
Hospital Virgen del Camino Pamplona

Dpt.Genética. Universidad Navarra Pamplona

VALENCIA:
Universitat d'Alicant. Alicante
Hospital La Fe. Valencia
Dpto. Patología. Universidad de Valencia.
Institut de Genètica Mèdica i Molecular (IGEM). C. Virgen del Consuelo Valencia

Annex 4. List of centres

ANDALUCIA:

Hospital Virgen del Rocío

Unit: Genética Médica y Diagnóstico Prenatal
Contact person: Guillermo Antiñolo Gil
Address: Avda. Manuel Siurot s/n. 41013 Sevilla
Telephone: 955 01 2778
E-mail: gantinolo@hvr.sas.junta-andalucia.es
Type of centre: public hospital

Hospital Virgen de la Macarena

Unit: Biología Molecular
Contact person: Miguel Lucas
Address: Avda Dr. Fedriani S/N 41071. Sevilla
Telephone: 955008114
E-mail: lucas@us.es
Type of centre: public hospital

Hospital San Cecilio

Unit: Bioquímica.
Contact person: José Antonio Gómez Capilla.
Address: Departamento de Bioquímica y Biología Molecular. Facultad de Medicina. Avd/ Madrid s/n. 18012. Granada.
Telephone: 958 243524/958023107/958243517
E-mail:jgomez@ugr.es
Type of centre: public hospital

Hospital Virgen de las Nieves

Unit: Laboratorio Genética.
Contact person: Matías Pérez
Address: Avda. Fuerzas Armadas.
13014 Granada
Telephone: 958020011
E-mail:mperez@hvn.sas.cica.es
Type of centre: public hospital

Hospital Carlos Haya

Unit: Servicio de hematología y hemoterapia
Contact person: Dr. Antonio Jimenez Velasco
Address: Avenida Carlos Haya s/n. 29010. Málaga
Telephone: 952645808
E-mail: jimeneza@hch.sas.junta-andalucia.es
Type of centre: public hospital

Unit: Laboratorio - Genética.
Arroyo de los Ángeles, s/n. 29011-Málaga
952304400. Dr. Marceliano Herránz. herranz@hch.sas.cica.es
Laboratorio. Inmunología: Antonio Alonso aalonso@hch.sas.junta-andalucia.es
Anatomía Patológica: Dolores Bautista. dbautista@hch.sas.junta-andalucia.es
952645838

NBT DIAGEN. CENTRO DE DIAGNOSTICO GENETICO
Contact person: Dr. Narain
Address: C/ Educardo Rivas 2-4. 41018 Sevilla
Telephone: 954575508
E-mail: narain@newbiotechnic.com
Type of centre: private laboratory

ARAGÓN:

Hospital Miguel Servet

Unit: Genética Médica
Contact person: Manuel Tamparillas Salvador
Address: Paseo Isabel La Católica 1 y 3. 50009 Zaragoza
Telephone: 976765500 Ext 3150
E-mail: mtamparilla@hmservet.insalud.es
Type of centre: public hospital

Centro de Analisis Genéticos C.A.G.T.

Unit: Genetica Clinica. Genética Molecular y Citogenética
Contact person: Dra. Ana Mª Palacio de Parada. Dra. Pilar Madero Barrajón
Address: C/ Teresa de Jesús 45-47. 50006- Zaragoza
Telephone: 976-55 64 84. Fax: 976- 30 69 10
E-mail: e-mail: cagt@arrakis.es
Type of centre: Private laboratory

Universidad de Zaragoza

Unit: Bioquímica y Biología Molecular
Contact person: Julio Montoya
Address: Miguel Servet 177
Telephone: +34-976761640
E-mail: jmontoya@posta.unizar.es
Type of centre: public university

Unit: Depart. Bioquimica, Biologia Molecular y Celular
Contact person: Miguel Pocoví Mieras
Address: Departamento Bioquímica, Biología Molecular y Celular. Fac. Ciencias. Universidad de Zaragoza. Campus Plaza San Francisco sn. 50009 Zaragoza
Telephone:34-976-761283
E-mail: mpocovi@posta.unizar.es
Type of centre: public university

ASTURIAS

Hospital Central de Asturias

Unit: Genética Molecular
Contact person: Dr. Eliecer Coto García
Celestino Villamil s/n 33006 Oviedo 33006 – OVIEDO – SPAIN
Telephone: 985.10.79.68
E-mail: ecoto@hcas.insalud.es
Type of centre: public hospital

BALEARES

Hospital Son Dureta. Mallorca

Unit: Genética
Contact person: Damian Heine Suñer
Address: Sección de Genética, H. Universitario Son Dureta. Avda. Andrea Doria 55. Palma de Mallorca 07014
Telephone: 971-175147
E-mail: dheine@hsd.es
Type of centre: public hospital

CANARIAS

Hospital Materno-Infantil de Canarias. Las Palmas

Unit: Laboratorio de Genética Molecular
Contact person: Dr. Miguel Fernández-Burriel Tercero
Address: Avda. Marítima del Sur s/n
35016-Las Palmas de Gran Canaria
Telephone: 928444790
E-mail: m.fernandez.012@recol.es
Type of centre: public hospital

Hospital Universitario de Canarias

Unit: Unidad de Investigación
Contact person: Eduardo Salido Ruiz
Address: Ofra-La Cuesta s/n. E-38320 Tenerife
Telephone: 922-319338
E-mail: esalido@ull.es
Type of centre: public hospital

CANTABRIA

Hospital Marqués de Valdecilla

Unit: Inmunología
Contact person: Francisco Leyva-Cobián
Address: Avenida de Valdecilla, s/n. 39008-Santander
Telephone: 942-202549
E-mail: fleyvaco@sarenet.es
Type of centre: public hospital

Unit: Neurología
Contact person: José Berciano
Telephone: 942-202520
E-mail: neuro@humv.es

CASTILLA LA MANCHA

Hospital Virgen de la Salud. Toledo

Unit: Genética
Contact person: Pedro Martínez
Address: Laboratorio de Genética. Hospital Nacional de Parapléjicos. Finca de la Peraleda s/n. 45071 Toledo
Telephone: 925-24 77 37
E-mail: pmartinez@cht.insalud.es
Type of centre: public hospital

CASTILLA-LEON

Instituto de Biología y Genética Molecular

Unit: Unidad de Diagnóstico Genético y Perinatal
Contact person: Juan José Tellería Orriols
Address: Avda. de Ramón y Cajal 7; 47005 Valladolid
Telephone: 983 423 189
E-mail: telleria@med.uva.es
Type of centre: public university

Universidad de Salamanca

Unit: Unidad de Medicina Molecular
Contact person: Rogelio González Sarmiento
Address: Facultad de Medicina. Avda. Campo Charro s/n 37007 Salamanca
Telephone: 9023294553
E-mail:gonzalez@usal.es
Type of centre: public university

CATALUÑA:

Hospital Clínico de Barcelona

Unit: Genética
Contact person: Dr. Rafael Oliva
Address: Villarroel 170, 08036 Barcelona
Telephone: 93 2275510
E-mail: oliva@medicina.ub.es

Unit: Centro de Diagnóstico Biomédico
Contact person: Josep Oriola
Telephone: 932275400
E-mail: joriola@clinic.ub.es
Type of centre: public hospital

Institut de Bioquímica Clínica. Clínic-Corporació Sanitària
Contact person : Dra. Teresa Pàmpols
Address : Institut de Bioquímica Clínica . Calle Mejía Lequerica s/n, Edificio Helios III, Planta baja . 08028 Barcelona
Telephone : 03 2275672
E-mail : tpampols@clinic.ub.es
Type of centre : public centre

H. la Santa Creu i Sant Pau

Unit: Servicio de Genética
Contact person: Montserrat Baiget
Address: Padre Claret 167. 08025 Barcelona
Telephone: 93-2919361
E-mail: mbaiget@hsp.santpau.es
Type of centre: public hospital

Hospital Valle Hebron

Unit: Centro de Investigacion en Bioquimica y Biologia Molecular
Contact person: Dr. Antonio Luis Andreu Periz
Address: P. Valle Hebron 119-129. 08035 Barcelona
Telephone: 934894057
E-mail: tandreu@hg.vhebron.es
Type of centre: public hospital

Unit: Unidad Investigación Endocrinología y Nutrición Pediátricas
Contact person: Dra. Laura Audi
Telephone: 934894030
E-mail: laudi@cs.vhebron.es

Institut Reserca Oncológica

Unit: Centro de Genética Médica y Molecular.
Personas de contacto: Virginia Nunes (Jefe de Departamento), V. Volpini(representante español en el EMQN)
Address: Gran vía s/n km 2,7. Hospitalet del Llobregat 08907, Barcelona
Telephone: 93 2607427
E-mail: vnunes@iro.es, vvolpini@iro.es
Type of centre: Research centre, private

Hospital Sant Joan de Déu, Barcelona

Unit: Sección Genética, Unidad Genética Molecular
Contact person: Eugenia Monrós
Address: Pss. Sant Joan de Déu 2, 08950 Esplugues
Telephone: 93 253 2103
E-mail: emonros@hsjdbcn.org
Type of centre: private hospital

Hospital del Mar - Facultad de Ciencias de la Salud (Universitat Pompeu Fabra)

Unit: Laboratorio de Citogenética y Biología Molecular (Dept. Patología) Unidad de Genética
Personas de contacto: Francesc Solé. Luis Pérez Jurado
Address: Paseo Marítimo 25-29. 08003 Barcelona
Telephone: 93-2483035
E-mail: e0037@imas.imim.es / luis.perez@cexs.upf.es
Type of centre: public hospital and public university

Corporación Parc Taulí. UDIAT-CD

Unit: Laboratorio, Genética
Contact person: Miriam Guitart
Address: Laboratorio. UDIAT-CD. Corporación Parc Taulí.
Parc Taulí s/n. 08208 Sabadell.(Barcelona)
Telephone:937231010 Ext.26111
E-mail: mguitart@cspt.es
Type of centre: public hospital

Laboratorio de Análisis Dr. Echevarne

Unit: Genética y Medicina Molecular
Contact person: Dr. José Ignacio Lao Villadóniga
Address: Provenza 312 (bajos). 08037 Barcelona
Telephone: 93-4964456
E-mail: jilao@echevarne.com
Type of centre: private laboratory

Centro de Patología Celular

Contact person: Dra. Marta Carrera
Address: C/ Londres, nº 6, 08029 Barcelona
Telephone: 93 322 88 06
E-mail: mca@sabater-tobella.com
Type of centre: private laboratory

EXTREMADURA:

Hospital Materno Infantil de Badajoz

Unit: Unidad de Genética
Contact person: Dr. Enrique Galan Gomez
Address: c/ Damián Téllez Lafuente s/n , Badajoz 06004
Telephone: 924-230400, EXT 287 (consulta), 458-459 (laboratorio)
E-mail: egala@ctv.es
Type of centre: public hospital

GALICIA:

Hospital de Conxo, FINGO

Unit: Unidad de Medicina Molecular
Contact person: Francisco Barros o Lourdes Loidi
Address: Hospital de Conxo, Rúa Ramón Baltar SN. 15706 Santiago de Compostela
Telephone: 981 951 888
E-mail: apimlbar@usc.es o lloidi@usc.es
Type of centre: public hospital

Centro Oncológico de Galicia.

Unit: laboratorio de Genética y Radiobiología
Contact person: A Mosquera
Address: Avda. Montserrat s/n 15009 A Coruña
Telephone: 981287499 ext 292
E-mail: genetica@cog.es
Type of centre: public hospital

Hospital Clinico Universitario de Santiago

Unit:Genética Médica
Contact person:Dra.A.Ansede
Address:C/Houpana s/n Santiago
Telephone:981950045
E-mail:branas@mixmail.comType of centre: Hospital Universitario Public
Type of centre: public hospital

H. Juan Canalejo

Unit: Hematologia y hemoterapia
Contact person: Dr. Javier Batlle Fonrodona.
Address: Edificio H. Teresa Herrera. Avda. Pasaje s/n. 15006 A CORUÑA
Telephone: 981 178000 (Ext 21113; 21115)
E-mail: jbatlle@canalejo.cesga.es
Type of centre: public hospital

S. Genética. Berta Rodríguez
Telephone: 981178000 berta_rodríguez@canalejo.org

MADRID:

Hospital Universitario La Paz

Unit: Unidad de Genética Molecular. Servicio de Bioquímica
Contact person: Jesús Molano Mateos
Telephone: 917277381
E-mail:jmolano@hulp.insalud.es
Address: Paseo de la Castellana 261; 28046 Madrid
Type of centre: public hospital

Unit: Unidad de Inmunología
Contact person: Mª Cruz García Rodríguez
Telephone: 91 7277095, 91 7277238
E-mail: mcruzgarcia@hulp.insalud.es

Hospital Gregorio Marañón

Unit: Servicio Bioquímica. Laboratorio de Diagnóstico Molecular.
Contact person: Dra Begoña Ezquieta
Address: c/ Doctor Esquerdo 46. 28007 Madrid
Telephone: 915868466
E-mail: bezquieta@airtel.net
Type of centre: public hospital

Fundación Jiménez Díaz. Madrid

Unit: Genética
Contact person: Carmen Ayuso // Carmen Ramos
Address: Av. Reyes católicos nº 2. 28040 Madrid
Telephone: 91/ 5446903 - 91/5504872
E-mail: Cayuso@fjd.es
Type of centre: private hospital

Hospital Clínico San Carlos

Unit: Genética
Contact person: Dra Lautre Ecenarro
Address: Ciudad Universitaria, s/n Madrid.
Telephone: 913303023
E-mail: mlautre@hcsc.insalud.es
Type of centre: public hospital

Unit: Imunología y Oncología Molecular
Contact person: Trinidad Caldés
Telephone: 913303348
E-mail: tcaldes@hcsc.es

Hospital 12 Octubre

Centro: Hospital Universitario 12 de Octubre
Unit: Centro de Investigación
Contact person: Joaquín Arenas
Address: Avda de Córdoba km 5.4. 28041 Madrid
Telephone: 91-390-8411
E-mail: jarenas@h120.es
Type of centre: public hospital

Hospital Ramón y Cajal

Centro: Hospital Ramón y Cajal
Unit: Unidad de Genética Molecular
Contact person: Felipe Moreno
Address: Carretera de Colmenar km 9,100. 28034 Madrid.
Telephone: 913368541
E-mail: fmoreno@hrc.insalud.es
Type of centre: public hospital

Centro de Diagnóstico de Enfermedades Moleculares

Contact person: Prof. M. Ugarte
Address: Dpto. Biología Molecular, Facultad de Ciencias, Universidad Autónoma de Madrid, 28049 Madrid
Telephone: 91 3974589
E-mail: cedem@cbm.uam.es
Type of centre: public university

Centro Nacional Investigaciones Oncológicas (CNIO)

Unit: Dpto Genética Humana
Contact person: Javier Benítez
Address: Melchor Fdez. Almagro 3 Madrid 28029
Telephone:912246965
E-mail:jbenitez@cnio.es
Type of centre: public research centre

Centro de Investigación sobre Anomalías Congénitas (CIAC), Instituto de Salud Carlos III. Ministerio de Sanidad y Consumo. Madrid.
Unit: Estudio Colaborativo Español de Malformaciones Congénitas
Contact person: Profa. Dra. María Luisa Martínez-Frías
Address: ISCIII. CIAC Calle, Sinesio Delgado, 6. Madrid
Telephone: 91 394 15 86
E-mail: luisama@med.ucm.es
Type of centre: public research centre

MURCIA

Centro de Bioquímica y Genética Clínica
Unit:Unidad de Genética Molecular
Contact person: Guillermo Glover
Address: Centro de Bioquímica y Genética Clínica. Labs Pabellon General. Hospital Virgen Arrixaca. 30120. El Palmar (Murcia)
Telephone:968 88 98 60
E-mail:guillermo.glover@carm.es
Type of centre: public centre

NAVARRA

Hospital Virgen del Camino Pamplona.

Unit: Genetica
Contact person: Alberto Valiente
Address: Irunlarrea,4; 31008; Pamplona
Telephone: 948/429991
E-mail: avalienm@cfnavarra.es
Type of centre: public hospital

Clínica Universitaria. Pamplona

Unit: Laboratorio de Biotecnología y Genómica
Contact person: Dr. Jesús García-Foncillas
Address: Avda Pio XII, 36. 31008-Pamplona
Telephone: 948296792/948296292.
E-mail: jgfoncillas@unav.es
Type of centre: private hospital

PAIS VALENCIA

Hospital Universitario La Fe. Valencia

Unit: Unidad de Genética y Diagnóstico Prenatal
Contact person: José Mª Millán Salvador
Address: Avda. Campanar, 21 Valencia 46009
Telephone: 96-3987370
E-mail: chema_millan@yahoo.es
Type of centre: public hospital

Facultad de Medicina Valencia

Unit: Departamento de Patologia
Contact person: Antonio Pellín Pérez
Address: Departamento de Patologia. Facultad de Medicina
Blasco Ibáñez, 17. Valencia 46010
Telephone: 969 3864145
E-mail: Antonio.Pellin@uv.es
Type of centre: public university

Facultad de Medicina Alicante. Universidad Miguel Hernández.
Unit: Unidad de Genética y Diagnóstico Prenatal. Departamento de Pediatría.
Contact person: Francisco Galan Sanchez
Address: Campus de San Juan. Apdo. de correos 18. 03550 – San Juan (Alicante)
Telephone: 96 – 5919433 ; Fax: 96 – 5919429
E-mail: fm.galan@umh.es
Type of centre: public university

Análisis Genéticos ANCOR S.L.

Contact person: Andrés Corno
Address: Avda Eusebio Sempere 22, 03003-Alicante
Telephone: 965 13 57 43-659 437 825 Fax :965 13 45 35
E-mail:ancor@vianwe.com
Type of centre: private laboratory

Institut de Genética Mèdica i Molecular, IGEM Valencia

Contact person: Javier García Planells
Address: Clínica Virgen del Consuelo. C/ Callosa d'Ensarrià, 12. Valencia 46007.
Telephone:96 317 78 00/ 96 317 78 21
E-mail: info@igem.es
Type of centre: private laboratory

PAIS VASCO

Hospital de Basurto

Unit: UNIDAD DE GENETICA (Laboratorio de Genética Molecular)
Contact person: Dra. Mª Isabel TEJADA
Address: Avda. Montevideo 18 / 48013-BILBAO
Telephone: 944006154
E-mail: itejada@hbas.osakidetza.net
Type of centre: public hospital

Hospital de Cruces

Unit: Unidad de Investigación
Contact person: Luis Castaño
Address: Plaza de Cruces S/N. 48903 Baracaldo. Vizcaya
Telephone: 946006376
E-mail: lcastano@hcru.osakidetza.net
Type of centre: public hospital

Hospital Donostia

Unit: Genética
Contact person: Dra. Ana María Cobo Esteban
Address: Paseo Dr Begiristain s/n San Sebastian 20014
Telephone: 943 007301
E-mail: anacobo@chdo.osakidetza.net
Type of centre: public hospital

Unit: Unidad Experimental
Contact person: Adolfo López de Munain
Telephone: 943 00 70 61
E-mail: uniexpe2@chdo.osakidetza.net
Type of centre: sanitario publico

Policlinica Gipuzkoa

Unit: Genética Molecular
Contact person: Dra. C. Vidales
Address: Paseo de Miramon, 174
20009- San Sebastián
Telephone: 943002837
E-mail: biomol@policlinicagipuzkoa.com
Type of centre: private hospital

RIOJA

Hospital San Millán. Logroño

Centro: Hospital "San Millán- San Pedro"
Unit: Laboratorio Central
Contact person: Luis Borque ó Fernando Iguaz
Address: C/ Autonomía de La Rioja, 3. 26004 Logroño
Telephone: 941-294500 Ext. 553 , 551, 2003
E-mail: lborque@arrakis.es y figuazp@nexo.es
Type of centre: public hospital

Annex 5. Conditions tested and centres arranged by functional areas

NEUROLOGIC CONDITIONS

	Centre	N. tests in 2001 (N/S when not specified by the centre)
Alzheimer disease (Presenilina 1, APP, APOE)	Hospital Clínico Barcelona	300
	H. Central de Asturias	220
	H Virgen del Camino	125
	U. Salamanca	50
	H. Valdecilla	25
	Centro de Bioquímica y Genética Clínica Murcia	APOE = 25
	H. Miguel Servet	23
	H. Universitario Tenerife	20
	H. Macarena Sevilla	20
	Clínica Universitaria Pamplona	15
	Laboratorio de Análisis Dr. Echevarne Barcelona	9
	Centro de Análisis Genéticos. Zaragoza	7
	H. Conxo. Santiago	3
	NBT DIAGEN Sevilla	N/S
	ANCOR Alicante	N/S
Amiliodean familiar polineuropathy	H. Son Dureta Mallorca	10
Aneuplodies by QFPCR	Laboratorio de Análisis Dr. Echevarne Barcelona	460
Angelman syndrome	Hospital Sant Joan de Déu, Barcelona	44
	H. La Fe. Valencia	35
	H. Miguel Servet	35
	F. Jiménez Díaz	25
	H. Son Dureta Mallorca	25
	H. Parc Tauli Barcelona	23
	H. Materno Infantil Badajoz	20
	H. Central de Asturias	13
	H Virgen del Camino	11
	Centro de Bioquímica y Genética Clínica Murcia	5
	Hospital Clínico Universitario Santiago	8
	Centro Oncológico de Galicia. A Coruña	4
	H. Virgen de la Salud Toledo	4
	IGEM Valencia	2
	H. del Mar – UPF Barcelona	2
	Laboratorio de Análisis Dr. Echevarne Barcelona	2
	H.Universitario Las Palmas	1
	Centro Investigación Anomalías Congénitas Madrid.	N/S
	H. Virgen de las Nieves. Granada	N/S
	H. V. Rocio Sevilla	N/S
CADASIL	Centro de Análisis Genéticos. Zaragoza	1
Charcot-Marie-Tooth neuropathy	IRO Barcelona	313
	Hospital Sant Joan de Déu, Barcelona	71
	H. La Fe. Valencia	53
	F. Jiménez Díaz	40
	H. Valdecilla	25
	Centro de Análisis Genéticos. Zaragoza	22
	IGEM Valencia	20
	Laboratorio de Análisis Dr. Echevarne Barcelona	14
	H. Conxo. Santiago	21
	Clínica Universitaria Pamplona	8
	H. Central de Asturias	6
Cryptic chromosomic alterations (FISH, Multitest-T and C)	Centro Investigación Anomalías Congénitas Madrid.	N/S
Dementia Fronto-temporal	Hospital Clínico Barcelona	30

	Clínica Universitaria Pamplona	6
Dentatorubral-pallidoluysian atrophy (DRPLA)	F. Jiménez Díaz	135
	H. La Fe. Valencia	37
	Hospital Clínico Universitario Santiago	5
	H. Central de Asturias	5
	Clínica Universitaria Pamplona	3
	Laboratorio de Análisis Dr. Echevarne Barcelona	2
	IRO Barcelona	1
	Hospital Clínico Barcelona	N/S
	H. Macarena Sevilla	N/S
	H. V. Rocio Sevilla	N/S
Dominant Ataxias (ADCAS) SCA1; SCA2 (Machado); SCA3; SCA6; SCA7; SCA8; SCA9; SCA10; SCA12	IRO Barcelona	203
	F. Jiménez Díaz	135
	Hospital Clínico Universitario Santiago	104
	H. Macarena Sevilla	N/S (all the ataxias 250)
	Laboratorio de Análisis Dr. Echevarne Barcelona	64
	H. La Fe. Valencia	37
	H. Conxo. Santiago	29
	H. Central de Asturias	25
	Centro de Análisis Genéticos. Zaragoza	13
	IGEM Valencia	10
	Hospital Clínico Barcelona	N/S
	Clínica Universitaria Pamplona	10
	H. V. Rocio Sevilla	N/S
Dominant macular dystrophies (RDS/periferina gene)	F. Jiménez Díaz	50
	H. V. Rocio Sevilla	N/S
Duchenne/Becker muscular dystrophy	H. San Pablo Barcelona	163
	H San Cecilio Granada	60
	F. Jiménez Díaz	45
	H. Universitario Las Palmas	26
	H. La Fe. Valencia	26
	Centro de Bioquímica y Genética Clínica Murcia	25
	H. La Paz. Madrid	20
	Hospital Sant Joan de Déu, Barcelona	14
	Laboratorio de Análisis Dr. Echevarne Barcelona	7
	Centro de Análisis Genéticos. Zaragoza	7
	Hospital Clínico Universitario Santiago	6
	IGEM Valencia	5
	Centro Oncológico de Galicia. A Coruña	4
	NBT DIAGEN Sevilla	N/S
	H. V. Rocio Sevilla	N/S
Facioscapulohumeral muscular dystrophy 1A (FSHMD1A)	H. Donostia. San Sebastián	100
	H. La Fe. Valencia	25
	IGEM Valencia	9
	H. V. Rocio Sevilla	N/S
Friedrich Ataxia (X25)	F. Jiménez Díaz	135
	Hospital San Juan de Dios, Barcelona	25
	Hospital Clínico Universitario Santiago	20
	H. Valdecilla	20
	H. La Fe. Valencia	19
	IRO Barcelona	9
	Laboratorio de Análisis Dr. Echevarne Barcelona	8
	Centro de Análisis Genéticos. Zaragoza	8
	H. Central de Asturias	7
	H. Conxo. Santiago	7
	IGEM Valencia	5

	Instituto Biología Genética Molecular. Valladolid	4
	Clínica Universitaria Pamplona	3
	H. Macarena Sevilla	N/S
	Hospital Clínico Barcelona	N/S
	H. Virgen de las Nieves. Granada	N/S
	H. V. Rocio Sevilla	N/S
Hirschsprung disease	Clínica Universitaria Pamplona	18
	H. Cruces Baracaldo	10
	H. V. Rocio Sevilla	N/S
Huntington disease	H. Macarena Sevilla	150
	F. Jiménez Díaz	85
	H. La Fe. Valencia	65
	H Virgen del Camino	50-60
	H. La Paz. Madrid	48
	H. Central de Asturias	15
	H. Conxo. Santiago	29
	H San Cecilio Granada	25
	Hospital Clínico Universitario Santiago	25
	Laboratorio de Análisis Dr. Echevarne Barcelona	20
	H. Son Dureta Mallorca	13
	Centro de Análisis Genéticos. Zaragoza	9
	H. Miguel Servet	4
	IGEM Valencia	3
	H. Virgen de las Nieves. Granada	N/S
	H. V. Rocio Sevilla	N/S
	Hospital Clínico Barcelona	N/S
	NBT DIAGEN Sevilla	N/S
Kallman syndrome (microdel Xp22)	F. Jiménez Díaz	6
	Centro de Análisis Genéticos. Zaragoza	11
	Laboratorio de Análisis Dr. Echevarne Barcelona	3
	H. V. Rocio Sevilla	N/S
Kennedy disease (AR, exón 1)	H. San Pablo Barcelona	6
	H. Macarena Sevilla	6
	Laboratorio de Análisis Dr. Echevarne Barcelona	4
	H. La Fe. Valencia	5
	H. Central de Asturias	5
	Clínica Universitaria Pamplona	3
	Instituto Biología Genética Molecular. Valladolid	1
	Centro de Análisis Genéticos. Zaragoza	1
	H. V. Rocio Sevilla	N/S
Leber optic atrophy	H. Macarena Sevilla	20
	Universidad Zaragoza. Dpto Biologia Molecular	20
	IRO Barcelona	19
	Clínica Universitaria Pamplona	5
	Laboratorio de Análisis Dr. Echevarne Barcelona	5
	Centro de Análisis Genéticos. Zaragoza	3
	H. Central de Asturias	2
	H. Conxo. Santiago	2
Lesh-Nyhan syndrome	H. La Paz. Madrid	2
Mental retardation FRAXE type linked (FMR2)	H. Basurto Bilbao	154
	H. La Fe. Valencia	100
	H.Universitario Las Palmas	93
	H Virgen del Camino	60
	Hospital Clínico Universitario Santiago	50
	Hospital Clínico Barcelona	N/S
	H. Son Dureta Mallorca	10
	F. Jiménez Díaz	2
Miller-Dieker syndrome	H. Son Dureta Mallorca	6
	H. V. Rocio Sevilla	N/S
Myotonic distrophy of waists (LGMD)	H. San Pablo Barcelona	25
Myotonic distrophy of waists (Type 2) (Calpainopathy, LGMD2A)	H. Donostia. San Sebastián	74
NARP	Laboratorio de Análisis Dr. Echevarne Barcelona	2
Neurofibromatosis type 1	IRO Barcelona	356

Neurofibromatosis (linkage análisis NF1 y NF2)	H. Ramón y Cajal	45
	IRO Barcelona	13 NF2
	Hospital Sant Joan de Déu, Barcelona	15
Neuropathy due to sensitivity to pressure	F. Jiménez Díaz	40
	Centro de Análisis Genéticos. Zaragoza	32
	H. Valdecilla	25
	Hospital Sant Joan de Déu, Barcelona	23
	H. Conxo. Santiago	11
	H. La Fe. Valencia	9
	IGEM Valencia	5
	Laboratorio de Análisis Dr. Echevarne Barcelona	3
Norrie disease (NPD)	F. Jiménez Díaz	8
	Centro de Análisis Genéticos. Zaragoza	1
Parkinson (Parkin, a-sinucleina)	H. Donostia. San Sebastián	26
	Hospital Clínico Barcelona	25
	H. Central de Asturias	18
	U. Salamanca	12
	Clínica Universitaria Pamplona	8
Prader Willi syndrome	Hospital Sant Joan de Déu, Barcelona	74
	H. La Fe. Valencia	60
	H. Materno Infantil Badajoz	40
	H. Parc Tauli Barcelona	30
	H. Son Dureta Mallorca	25
	F. Jiménez Díaz	25
	Centro de Bioquímica y Genética Clínica Murcia	15
	H. Central de Asturias	13
	H Virgen del Camino	11
	Hospital Clínico Universitario Santiago	10
	H.Universitario Las Palmas	9
	Laboratorio de Análisis Dr. Echevarne Barcelona	7
	H. Virgen de la Salud Toledo	6
	H. Miguel Servet	6
	Centro Oncológico de Galicia. A Coruña	5
	IGEM Valencia	4
	H. del Mar – UPF Barcelona	3
	Centro de Análisis Genéticos. Zaragoza	2
	Centro Investigación Anomalías Congénitas Madrid.	N/S
	H. Virgen de las Nieves. Granada	N/S
	NBT DIAGEN Sevilla	N/S
	H. V. Rocio Sevilla	N/S
Rett syndrome	Hospital Sant Joan de Déu, Barcelona	244
	H. Conxo. Santiago	3
	H. V. Rocio Sevilla	N/S
Schizophrenia	H. Valdecilla	102
Silver-Russel syndrome	H. Son Dureta Mallorca	5
	Hospital Clínico Barcelona	N/S
Smith-Magenis syndrome	H. Son Dureta Mallorca	10
	H. Materno Infantil Badajoz	8
	Hospital Clínico Universitario Santiago	5
	H. del Mar – UPF Barcelona	3
	H. Conxo. Santiago	1
	H. V. Rocio Sevilla	N/S
	F. Jiménez Díaz	N/S
Spinal muscular atrophy (Werdnig-Hoffmann y Kugelberg-Welander) (SMN)	H. San Pablo Barcelona	298
	H. Universitario Las Palmas	34
	H. Ramón y Cajal	127
	H. La Fe. Valencia	14
	Centro de Análisis Genéticos. Zaragoza	11
	Laboratorio de Análisis Dr. Echevarne Barcelona	8
	IGEM Valencia	4
	Instituto Biología Genética Molecular. Valladolid	2

	H. Macarena Sevilla	N/S
	H. V. Rocio Sevilla	N/S
Steinert Myotonic dystrophy	H. Macarena Sevilla	300
	H. San Pablo Barcelona	263
	H. La Paz. Madrid	80
	F. Jiménez Díaz	80
	H. La Fe. Valencia	66
	H. Donostia. San Sebastián	44
	Centro de Bioquímica y Genética Clínica Murcia	35
	H Virgen del Camino Pamplona	25
	H. Cruces Baracaldo	25
	H. Miguel Servet	22
	H. Valdecilla	20
	H. Universitario Las Palmas	19
	H. Basurto Bilbao	15
	Centro de Análisis Genéticos. Zaragoza	15
	Hospital Sant Joan de Déu, Barcelona	14
	Laboratorio de Análisis Dr. Echevarne Barcelona	12
	Hospital Clínico Universitario Santiago	10
	H. Central de Asturias	6
	IGEM Valencia	4
	H. Virgen de las Nieves. Granada	N/S
	NBT DIAGEN Sevilla	N/S
	H. V. Rocio Sevilla	N/S
Torsion dystonia (DYT1)	F. Jiménez Díaz	20
	Clínica Universitaria Pamplona	15
	H. Son Dureta Mallorca	4
	H. La Fe. Valencia	4
	Centro de Análisis Genéticos. Zaragoza	4
	IGEM Valencia	1
Tuberose sclerosis	H. Valdecilla	N/S
Williams-Beuren syndrome (LOH , FISH) (ATP7B)	H. del Mar – UPF Barcelona	30
	Hospital Sant Joan de Déu, Barcelona	26
	H. La Fe. Valencia (loh)	24
	H. Materno Infantil Badajoz	15
	H. Son Dureta Mallorca	15
	H.Universitario Las Palmas	14
	Hospital Clínico Universitario Santiago	8
	H. Central de Asturias	5
	H. Parc Tauli Barcelona	5
	F. Jiménez Díaz	5
	H. Virgen de la Salud Toledo	5
	IGEM Valencia	2
	H. Conxo. Santiago	2
	Hospital Clínico Barcelona	N/S
	Centro Investigación Anomalías Congénitas Madrid.	N/S
	H. Virgen de las Nieves. Granada	N/S
	H. V. Rocio Sevilla	N/S
Wolf-Hirshhorm syndrome	H. V. Rocio Sevilla	N/S
Wolfram syndrome	IRO Barcelona	8
X-Frágil syndrome (FMR1)	Hospital Sant Joan de Déu, Barcelona	308
	H. La Fe. Valencia	295
	H. Basurto Bilbao	268
	Centro de Bioquímica y Genética Clínica Murcia	250
	H. Macarena Sevilla	225
	H. Ramón y Cajal	195
	H. Son Dureta Mallorca	170
	H. Miguel Servet	129
	F. Jiménez Díaz	110
	H.Universitario Las Palmas	93
	H San Cecilio Granada	90
	Centro Oncológico de Galicia. A Coruña	80
	Centro de Análisis Genéticos. Zaragoza	73
	H. Virgen de la Salud Toledo	63

H Virgen del Camino	60
Instituto Biología Genética Molecular. Valladolid	53
IGEM Valencia	50
Laboratorio de Análisis Dr. Echevarne Barcelona	39
Hospital Clínico Universitario Santiago	35
H. Central de Asturias	17
H. del Mar – UPF Barcelona	10
Hospital Clínico Barcelona	N/S
ANCOR Alicante	N/S
H. Virgen de las Nieves. Granada	N/S
H. V. Rocio Sevilla	N/S

HEMATOLOGIC CONDITIONS

	Centre	N. tests 2001
ACE Angiotensin I convertor enzyme	NBT DIAGEN Sevilla	N/S
Bruton type inmuno-deficiency	H. La Fe. Valencia	14
Chimerism determination for bone marrow transplants	H. Carlos Haya	221
	H. Son Dureta Mallorca	60
	Centro Oncológico de Galicia. A Coruña	6
	H. Virgen de las Nieves. Granada	N/S
Drepanocitosis	H. San Pablo Barcelona	5
Factor II G20210A	NBT DIAGEN Sevilla	N/S
Haemophilia A	H. San Pablo Barcelona	113
	Laboratorio de Análisis Dr. Echevarne Barcelona	3
	IGEM Valencia	2
	ANCOR Alicante	N/S
Haemophilia B	H. San Pablo Barcelona	22
	Laboratorio de Análisis Dr. Echevarne Barcelona	1
	ANCOR Alicante	N/S
Leukemia	H. del Mar – UPF Barcelona	800
	H. Conxo. Santiago	168
	H. Carlos Haya	147
	CNIO Madrid	100
	H Virgen del Camino	50
	Laboratorio de Análisis Dr. Echevarne Barcelona	16
	ANCOR Alicante	N/S
	H. Virgen de las Nieves. Granada	N/S
Leukemia and limphoma	H. Virgen de la Salud Toledo	375 (mol) 190 (citog)
	NBT DIAGEN Sevilla	N/S
Leukemia and limphoma (translocations)	Centro Oncológico de Galicia. A Coruña	225
Metilen TetraHidrofolato Reductasa (MTHFR)	H Virgen del Camino Pamplona	250
	H. Gregorio Marañón	N/S
	NBT DIAGEN Sevilla	N/S
Mut C>T 677 MTHFR gene	H Virgen del Camino Pamplona	250
	H. Conxo. Santiago	50
Other oncohematologic conditions	CNIO Madrid	300
PAI-I Plasminogen activator inhibitor	NBT DIAGEN Sevilla	N/S
Sideroblastic familiar anemia X-linked	H. La Paz. Madrid	2
Talasemia Alpha	H. San Pablo Barcelona	20
	Centro de Análisis Genéticos. Zaragoza	11
	Laboratorio de Análisis Dr. Echevarne Barcelona	9
	H. Virgen de las Nieves. Granada	N/S
Talasemia Beta	H. San Pablo Barcelona	46
	Centro de Análisis Genéticos. Zaragoza	4
	Laboratorio de Análisis Dr. Echevarne Barcelona	3
	H. Virgen de las Nieves. Granada	N/S
Thrombophilia (Mut. G1691A factor V Leiden gene) (Mut G20210A protrombina gene)	Hospital Juan Canalejo	2160
	H. Conxo. Santiago	261
	Laboratorio de Análisis Dr. Echevarne Barcelona	252
	H Virgen del Camino Pamplona	250
	Centro de Análisis Genéticos. Zaragoza	230
	H. Carlos Haya	130 N/S
	H. Central de Asturias.	127
	H. Universitario Tenerife	50

	IRO Barcelona	85
	U. Salamanca	50
	F. Jiménez Díaz	35
	H. Valdecilla	14
	ANCOR Alicante	N/S
	H. Gregorio Marañón	N/S
	H. V. Rocio Sevilla	N/S
Thrombophilia (Mut C677T)	H. Valdecilla	14
Thrombophilia (metilentetrahidrofolatoreductasa)	H. Valdecilla	14
Von Willebrand disease	Hospital Juan Canalejo	400

NEOPLASIC CONDITIONS

	Centre	N. tests 2001
Adenomatous polyposis of the colon (APC)	Clínica Universitaria Pamplona	36
	H Virgen del Camino Pamplona	28
	H. La Fe. Valencia	20
	H. Conxo. Santiago	19
	CNIO Madrid	10
	Laboratorio de Análisis Dr. Echevarne Barcelona	7
	H. Valdecilla	5
	Centro de Análisis Genéticos. Zaragoza	1
	H. San Carlos Madrid	N/S
	Hospital Clínico Barcelona	N/S
	H. Virgen de las Nieves. Granada	N/S
	NBT DIAGEN Sevilla	N/S
	H. V. Rocio Sevilla	N/S
Adenomatous polyposis of the colon (Gen P53, Gen kRAS, Gen hMHS2, Gen hMLH1, Gen hPMS1,Gen hPMS2)	Clínica Universitaria Pamplona	37
	H. Valdecilla	5 each mutation
Beta tubulina	H. Miguel Servet	10
Bladder sporadic cancer	U. Valencia	40
Breast cancer (BCRA1 y BCRA2)	H. San Pablo Barcelona	79
	H. San Carlos Madrid	50
	U. Salamanca	50
	Clínica Universitaria Pamplona	50
	CNIO Madrid	45
	H. del Mar – UPF Barcelona	20
	H. Conxo. Santiago	13
	Laboratorio de Análisis Dr. Echevarne Barcelona	2
	Centro de Análisis Genéticos. Zaragoza	2
	Hospital Clínico Barcelona	N/S
	H. V. Rocio Sevilla	N/S
	ANCOR Alicante	N/S
CK-19	Clínica Universitaria Pamplona	178
	U. Salamanca	12
Familiar feochromocitome	CNIO Madrid	5
	H. Cruces Baracaldo	3
	Clínica Universitaria Pamplona	3
	Centro de Análisis Genéticos. Zaragoza	1
	H. La Fe. Valencia	N/S
	NBT DIAGEN Sevilla	N/S
	H. V. Rocio Sevilla	N/S
Familiar medullar thyroid cancer FMTC	H. V. Rocio Sevilla	N/S
	Hospital Clínico Barcelona	N/S
Familiar melanoma (pl5, p19,CDK4)	Clínica Universitaria Pamplona	12
	Hospital Clínico Barcelona	N/S
	Centro de Análisis Genéticos. Zaragoza	2
Familiar melanoma (pl6)	Clínica Universitaria Pamplona	12

	CNIO Madrid	5
GST mu (colon, bladder)	U. Salamanca	400
	Clínica Universitaria Pamplona	59
HLXB9	CNIO Madrid	2
HNPCC markers for MSI Panel 5 micro satellites instability	ANCOR Alicante	N/S
K-RAS (CODON 12, 13) (lung, colon, ovary, larynx)	Clínica Universitaria Pamplona	185
	H. Cruces Baracaldo	50
	U. Salamanca	25
	ANCOR Alicante	N/S
Kidney sporadic cancer	U. Valencia	60
Li-Fraumeni disease (p53)	U. Salamanca	10
	Clínica Universitaria Pamplona	9
	H. San Pablo Barcelona	8
	Laboratorio de Análisis Dr. Echevarne Barcelona	3
	H. Conxo. Santiago	2
	Centro de Análisis Genéticos. Zaragoza	2
	H. San Carlos Madrid	N/S
	ANCOR Alicante	N/S
	Centro Oncológico de Galicia. A Coruña	N/S
	NBT DIAGEN Sevilla	N/S
	H. V. Rocio Sevilla	N/S
Lynch Syndrome	Centro de Bioquímica y Genética Clínica Murcia	15
	H. V. Rocio Sevilla	N/S
	NBT DIAGEN Sevilla	N/S
	H. Virgen de las Nieves. Granada	N/S
Meningions sporadic cancer	U. Valencia	20
Multiple Endocrine Neoplasia type 1 (MEN1)	H. Cruces Baracaldo	40
	Centro de Bioquímica y Genética Clínica Murcia	25
	CNIO Madrid	20
	Hospital Clínico Barcelona	N/S
	Clínica Universitaria Pamplona	7
Multiple Endocrine Neoplasia, type IIA (MEN2A)	CNIO Madrid	40
	Centro de Bioquímica y Genética Clínica Murcia	25
	Clínica Universitaria Pamplona	19
	H. La Fe. Valencia	16
	H. Cruces Baracaldo	5
	H. Conxo. Santiago	4
	Centro de Análisis Genéticos. Zaragoza	2
	H. Virgen de las Nieves. Granada	N/S
	NBT DIAGEN Sevilla	N/S
	H. V. Rocio Sevilla	N/S
Multiple Endocrine Neoplasia type II B (MEN2B)	H. Cruces Baracaldo	5
	Centro de Análisis Genéticos. Zaragoza	2
	H. La Fe. Valencia.	N/S
	H. Conxo. Santiago	N/S
	NBT DIAGEN Sevilla	N/S
	H. V. Rocio Sevilla	N/S
Neuroblastomes sporadic cancer	U. Valencia	70
Non polyposic colonic cancer	CNIO Madrid	15
	Hospital Clínico Barcelona	N/S
p53 EXONES 4,5,6,7,8 (Lung, breast, ovary, Endomet.)	Clínica Universitaria Pamplona	289
	U. Salamanca	200
	H. San Carlos	130
	H. Miguel Servet	105
	H. Cruces Baracaldo	50
	U. Valencia	50
	H. Conxo. Santiago	47
	CNIO Madrid	10
	Laboratorio de Análisis Dr. Echevarne Barcelona	5
	Centro de Análisis Genéticos. Zaragoza	2

	ANCOR Alicante	N/S
	H. V. Rocio Sevilla	N/S
	H. Macarena Sevilla	N/S
PSA (prostate cancer)	Clínica Universitaria Pamplona	150
PTEN	CNIO Madrid	5
Retinoblastoma	H. La Fe. Valencia	26
	Centro de Análisis Genéticos. Zaragoza	1
Sarcomes sporadic cancer	U. Valencia	50
Thyroid medullar sporadic cancer	H. Cruces Baracaldo	15
	H. V. Rocio Sevilla	N/S
Tirosinasa (tyroid cancer)	H. Conxo. Santiago	43
Tirosinasa (melanoma)	H. Conxo. Santiago	16
Von Hippel Lindau disease	CNIO Madrid	30
	Clínica Universitaria Pamplona	12
	H. La Fe. Valencia	8
	U. Salamanca	5
	Centro de Análisis Genéticos. Zaragoza	2
	H. Conxo. Santiago	1
	Hospital Clínico Barcelona	N/S
	H. V. Rocio Sevilla	N/S

METABOLIC CONDITIONS

	Centre	N. tests 2001
3-beta hydroxiesteroid-deshydrogenase deficiency	H. Conxo. Santiago	1
5 a-reductasa tipo 2 deficiency	H. Valle Hebrón Barcelona	5
11-beta-hidroxilasa deficiency	H. Gregorio Marañón	4
	H. Conxo. Santiago	4
	H. San Millán. Logroño	N/S
17-alpha-hydroxylase deficiency	H. Conxo. Santiago	2
	H. San Millán. Logroño	N/S
17-beta hydroxiesteroid-deshydrogenase deficiency	H. de Cruces Baracaldo	2
	H. Valle Hebrón	2
21-hydroxylase deficiency	H. Gregorio Marañón Madrid	400
	H. La Paz. Madrid	352
	H. Conxo. Santiago	60
	Hospital Sant Joan de Déu, Barcelona	51
	Laboratorio de Análisis Dr. Echevarne Barcelona	3
	H. San Millán. Logroño	N/S
Adrenal hyperplasia congenital	H. La Paz. Madrid	352
	H. Conxo. Santiago	64
	Hospital Clínico Barcelona	N/S
	H. Central de Asturias.	17
Adrenal hypoplasia congenital	H. Conxo. Santiago	1
Alfa-1-antitripsina deficiency	H. La Paz. Madrid	54
	H. Macarena Sevilla	50
	NBT DIAGEN Sevilla	N/S
	H. San Millán. Logroño	N/S
Apolipoprotein B (APOB) deficiency	Universidad Zaragoza. Dpto Biologia Molecular	930
	H. Conxo. Santiago	2
	NBT DIAGEN Sevilla	N/S
Carnitina palmitoil transferasa II deficiency (Y628S mutation)	H. 12 de Octubre	148
Carnitina palmitoil transferasa II deficiency (S113L mutation)	H. 12 de Octubre	20
	Centro de Diagnóstico de Enfermedades Moleculares Madrid	1
Cerebrotendinosis Xantomatosa (CYP27)	H. Conxo. Santiago	1
Maturity-onset diabetes of the	H. La Paz. Madrid	76

young MODY type (GK, HNF1)		
	H. de Cruces Baracaldo	25
	Hospital Clínico Barcelona	N/S
Diabetes type I (HLA susceptibility)	H. La Paz. Madrid	24
Gaucher disease	Universidad Zaragoza. Dpto Biologia Molecular	68
	Hospital Clínico Barcelona	N/S
Growth hormone deficiency	H. Valle Hebrón Barcelona	> 100
	H. Conxo. Santiago	43
	H. La Paz. Madrid	8
	Hospital Clínico Barcelona	N/S
	H. San Millán. Logroño	N/S
Growth hormone insensibility	H. Conxo. Santiago	71
	H. La Paz. Madrid	N/S
Hurler disease	Laboratorio de Análisis Dr. Echevarne Barcelona	1
	Hospital Clínico Barcelona	N/S
Hunter disease	H. La Fe. Valencia	2
	Hospital Clínico Barcelona	N/S
Hypercholesterolemia	Universidad Zaragoza. Dpto Biologia Molecular	350
	H. Conxo. Santiago	2
Hyperlipoproteinemia type III, disbetalipoproteinemia	Universidad Zaragoza. Dpto Biologia Molecular	25
McArdle disease (miofosforilasa deficiency) Mutations R49X, G204S y W797R	H. 12 de Octubre	134
Medium Chain Acil-CoA deshidrogenasa deficiency (MCAD) (K304E mutation)	Laboratorio de Análisis Dr. Echevarne Barcelona	16
	Instituto Biología Genética Molecular. Valladolid	15
	Hospital Clínico Barcelona	15
	H. Conxo. Santiago	6
	H. Universitario Las Palmas	5
	Centro de Diagnóstico de Enfermedades Moleculares Madrid	2
	H. Gregorio Marañón	N/S
	Hospital Clínico Barcelona	N/S
	H. V. Rocio Sevilla	N/S
Methyl crotonil glicinuria (MCCA and MCCB Genes)	Centro de Diagnóstico de Enfermedades Moleculares Madrid	16
Methylenetetrahydrofolate-5,10 reductase (MTHFR) 677C→T	Centro de Diagnóstico de Enfermedades Moleculares Madrid	22
MTRR 66 G→A	Centro de Diagnóstico de Enfermedades Moleculares Madrid	22
Mioadenilato desaminasa deficiency, Q12X mutation	H. 12 de Octubre	36
Mucolipidosis III	Centro de Análisis Genéticos. Zaragoza	1
Partial familiar lipodistrophy	H. Conxo. Santiago	1
Phenylketonuria- Phenylalaninemia (PAH)	Centro de Diagnóstico de Enfermedades Moleculares Madrid	45
Primary hyperaldosteronism	H. Conxo. Santiago	5
	H. Universitario Tenerife	2
	H. San Millán. Logroño	N/S
Primary hyperoxaluria	H. Universitario Tenerife	20
Propionic acidemia	Centro de Diagnóstico de Enfermedades Moleculares Madrid	25
Propionic acidemia (PCCA and PCCB Genes)	Centro de Diagnóstico de Enfermedades Moleculares Madrid	25
Smitz-Lemli-Opitz syndrome 7-Esterol Reductasa (DRCR7)	Centro de Análisis Genéticos. Zaragoza	1
	Hospital Clínico Barcelona	N/S
Triple A syndrome	H. Conxo. Santiago	3

PRIMARY INMUNODEFICIENCIES

	Centre	N. tests 2001
Autosomic agammaglobulinemia (μ chain)	H. La Paz. Madrid	4 families
Agammaglobulinemia X linked	H. La Paz. Madrid	54 families
Apoptosis deficiency (APO 1)	H. La Paz. Madrid	N/S
B. Cesar-Chediak-Higashi disease	H. Valdecilla	N/S
Combined deficiency X linked (γR-IL2 chain)	H. La Paz. Madrid	5 families
Complement deficiencies	H. Valdecilla	N/S
Di George syndrome	H. Valdecilla	4
HLA type II molecules deficiency	H. Valdecilla	N/S
Hyper IgM syndrome	H. La Paz. Madrid	6 families
Leukocyte adhesion deficiency, type I and II	H. Valdecilla	N/S
Omenn syndrome, RAG 1, RAG 2 Combined autosomic recesive deficiency	H. La Paz. Madrid	5 families
	H. Valdecilla	1
Screening for poligenetic disorders	H. Valdecilla	>200
Wisckott-Aldrich syndrome	H. Valdecilla	N/S

OTHER CONDITIONS

	Centre	N. tests 2001
Absence of deferent conduct	NBT DIAGEN Sevilla	N/S
Achondroplasia/Hipochondroplasia (FGFR3)	H. Gregorio Marañón Madrid	50
	H. La Paz. Madrid	44
	F. Jiménez Díaz	20
	H. Son Dureta Mallorca	5
	H. Conxo. Santiago	3
	H. Central de Asturias.	3
	Centro de Bioquímica y Genética Clínica Murcia	2
	H.Universitario Las Palmas	2
	Centro de Análisis Genéticos. Zaragoza	2
	Hospital Clínico Barcelona	N/S
	H. Miguel Servet	2
Alport syndrome	H. Conxo. Santiago	2
AIDS (□32-CCR5, V64I-CCR2, SDF1-3´A, RANTES-403C>A, RANTES-28C>G)	H. V. Rocio Sevilla	N/S
Aneuploidies	Centro Oncológico de Galicia. A Coruña	80
Azoospermia and oligospermia (Microdelection cr. Y)	Hospital Clínico Barcelona	290
	H. Virgen de la Salud Toledo	86
	Centro de Patología Celular. Barcelona	61
	H. Son Dureta Mallorca	50
	H. Macarena Sevilla	50
	F. Jiménez Díaz	40
	Laboratorio de Análisis Dr. Echevarne Barcelona	39
	H. Miguel Servet	36
	U. Salamanca	25
	H. Universitario Tenerife	24
	H. Central de Asturias.	21
	Centro de Bioquímica y Genética Clínica Murcia	10
	Centro de Análisis Genéticos. Zaragoza	13
	Centro Oncológico de Galicia. A Coruña	10
	H Virgen del Camino	8
	Hospital Clínico Universitario Santiago	3
	NBT DIAGEN Sevilla	N/S
Beckwith-Wiedemann syndrome	H. La Fe. Valencia	6

Cavernomatosis	H. Macarena Sevilla	140
Celiac disease	H Virgen del Camino Pamplona	250
	Instituto Biología Genética Molecular. Valladolid	150
	Laboratorio de Análisis Dr. Echevarne Barcelona	21
	IGEM Valencia	4
	H. San Millán. Logroño	N/S
Celiac disease (HLA)	Policlinica Gipuzkoa. San Sebastián	25
Cistinuria	IRO Barcelona	20
Coroideremia	F. Jiménez Díaz	10
Cystic fibrosis	IRO Barcelona	574
	Instituto Biología Genética Molecular. Valladolid	200
	H. Macarena Sevilla	180
	H. La Fe. Valencia	179
	H. Son Dureta Mallorca	110
	H. La Paz. Madrid	93
	Centro Oncológico de Galicia. A Coruña	60
	H. Ramón y Cajal	53
	Centro de Bioquímica y Genética Clínica Murcia	50
	F. Jiménez Díaz	50
	Centro de Patología Celular. Barcelona	44
	H.Universitario Las Palmas	43
	H Virgen del Camino	43
	Centro de Análisis Genéticos. Zaragoza	36
	H. Cruces Baracaldo	32
	Hospital Sant Joan de Déu, Barcelona	31
	H San Cecilio Granada	30
	Hospital Clínico Universitario Santiago	23
	H. Miguel Servet	19
	H. Universitario Tenerife	18
	Laboratorio de Análisis Dr. Echevarne Barcelona	13
	H. Central de Asturias.	9
	H. Conxo. Santiago	5
	IGEM Valencia	5
	H. Gregorio Marañón Madrid	N/S
	H. Virgen de las Nieves. Granada	N/S
	Hospital Clínico Barcelona	N/S
	NBT DIAGEN Sevilla	N/S
	H. V. Rocio Sevilla	N/S
Deafness, linked to Conexina 26 gene	H. Ramón y Cajal	500
	Clínica Universitaria Pamplona	156
	H.Universitario Las Palmas	31
	F. Jiménez Díaz	5
	Centro de Análisis Genéticos. Zaragoza	1
	H. V. Rocio Sevilla	N/S
Deafness, linked to Conexina 32 gene	Centro de Análisis Genéticos. Zaragoza	2
Deafness (mitochondrial)	H. Ramón y Cajal	600
Deafness, neurosensorial, linked to mitochondrial DNA, A1555G mutation	H. 12 de Octubre	36
Deafness, mitochondrial (by ototoxics)	F. Jiménez Díaz	4
Deafness sporadic (otoferlina gene)	H. Ramón y Cajal	300
Di George syndrome	H. La Fe. Valencia	84
	Hospital Clínico Universitario Santiago	40
	H. Son Dureta Mallorca	35
	Centro Oncológico de Galicia. A Coruña	30
	H. Miguel Servet	15 (FISH)
	H Virgen del Camino	11
	Centro de Patología Celular. Barcelona	9
	IGEM Valencia	6
	H. Conxo. Santiago	6
	H. V. Rocio Sevilla	N/S
	F. Jiménez Díaz	N/S
Di George syndrome / S. Velocardiofacial syndrome (FISH)	H. Parc Tauli	32
Emery-Dreifuss disease	H. La Fe. Valencia	3

Epidermolysis bullosa	H. La Fe. Valencia	1
Espondiloartropaties (b27 genotyping)	Policlinica Gipuzkoa. San Sebastián	N/S
Factor V Deficiency	H. Miguel Servet	1241
	H. Cruces Baracaldo	600
	Centro de Análisis Genéticos. Zaragoza	222
	Laboratorio de Análisis Dr. Echevarne Barcelona	134
	H. Central de Asturias.	127
	H. Carlos Haya	67
	IRO Barcelona	58
	U. Salamanca	50
	H. Conxo. Santiago	43
	F. Jiménez Díaz	35
	H. Universitario Tenerife	20
	ANCOR Alicante	N/S
	H. Gregorio Marañón Madrid	N/S
	H. Virgen de las Nieves. Granada	N/S
	NBT DIAGEN Sevilla	N/S
	H. V. Rocio Sevilla	N/S
Familiar hipercolesterolemia and SRY gene	U. Alicante	N/S
Gilbert syndrome	H. San Pablo Barcelona	190
Hemocrhomatosis (HFE)	H. Cruces Baracaldo	968
	H. Miguel Servet	410
	H. San Pablo Barcelona	385
	H. La Fe. Valencia	314
	Hospital Clínico Barcelona	300
	Centro de Análisis Genéticos. Zaragoza	312
	Laboratorio de Análisis Dr. Echevarne Barcelona	257
	F. Jiménez Díaz	207
	H. Macarena Sevilla	200
	H. Conxo. Santiago	187
	H Virgen del Camino	162
	H. La Paz. Madrid	162
	H. San Millán. Logroño	110
	U. Valencia	100
	U. Salamanca	75
	H. Universitario Tenerife	64
	H San Cecilio Granada	60
	Instituto Biología Genética Molecular. Valladolid	60
	Centro Oncológico de Galicia. A Coruña	30
	H. Son Dureta Mallorca	20
	Hospital Clínico Universitario Santiago	10
	IGEM Valencia	5
	H. V. Rocio Sevilla	N/S
	H. Virgen de las Nieves. Granada	N/S
	NBT DIAGEN Sevilla	N/S
	ANCOR Alicante	N/S
Hemochromatosis (MUTACIONES C282Y, H63D, S65C)	Policlinica Gipuzkoa. San Sebastián	15
Holoprosencefalia	H. Conxo. Santiago	1
Homocystinuria MTHFR	H. Miguel Servet	1241
	Hospital Sant Joan de Déu, Barcelona	81
	Laboratorio de Análisis Dr. Echevarne Barcelona	31
	IRO Barcelona	1
	H. Conxo. Santiago	1
	H. Virgen de las Nieves. Granada	N/S
HPV	Laboratorio de Análisis Dr. Echevarne Barcelona	295
Incontinentia pigmenti	H. La Fe. Valencia	N/S
Mediterranean familiar fever	F. Jiménez Díaz	20
	H. Valdecilla	17
	Hospital Clínico Universitario Santiago	2
	Centro de Análisis Genéticos. Zaragoza	2

	H. V. Rocio Sevilla	N/S
Mental retardation familiar inespecific	H. La Fe. Valencia	21
Miller-Diecker syndrome	H. La Fe. Valencia	3
Morris syndrome, Reifenstein syndrome (AR exons 2-8)	H. Valle Hebrón	30
	H. Cruces Baracaldo	5
	Clínica Universitaria Pamplona	5
	H. Conxo. Santiago	3
	Hospital Clínico Barcelona	N/S
Narcolepsy (HLA markers)	Policlinica Gipuzkoa. San Sebastián	N/S
Nephronophthisis, familial juvenile 1(NPHP1)	H. Central de Asturias.	3
	H. Universitario Tenerife	2
	Hospital Clínico Barcelona	N/S
Non Hodgking lynphomes: bcl2/IgH y Bcl1/IgH	H. Carlos Haya	68
Norrie disease (NDP)	F. Jiménez Díaz	8
	Centro de Análisis Genéticos. Zaragoza	1
	Hospital Clínico Barcelona	N/S
NPC1	H. Conxo. Santiago	N/S
Panhipopituitarism	H. Conxo. Santiago	11
	H. Cruces Baracaldo	10
Poliendrocrinopathy autoinmmune type 1	H. Conxo. Santiago	1
Polycystic Kidney Disease I (PKD1)	H. Conxo. Santiago	14
	H. Ramón y Cajal	37
	H. Central de Asturias.	13
	Centro de Análisis Genéticos. Zaragoza	4
	Hospital Clínico Barcelona	N/S
	H. Son Dureta Mallorca	N/S
Polycystic Kidney Disease 2 (PKD2)	H. Central de Asturias.	7
	H. Conxo. Santiago	2
	H. Son Dureta Mallorca	N/S
	Hospital Clínico Barcelona	N/S
Porphyria Acute intermittent (AIP)	H. La Paz. Madrid	29
Porphyria, congenital erythropoietic	H. La Paz. Madrid	N/S
Protrombine gene	H. Miguel Servet	1241
	H. Cruces Baracaldo	600
	Centro de Análisis Genéticos. Zaragoza	236
	H. Conxo. Santiago	218
	H. Central de Asturias.	127
	Laboratorio de Análisis Dr. Echevarne Barcelona	114
	H. Carlos Haya	63
	IRO Barcelona	52
	U. Salamanca	50
	F. Jiménez Díaz	35
	H. Universitario Tenerife	20
	H. Gregorio Marañón Madrid	N/S
	H. Virgen de las Nieves. Granada	N/S
	H. V. Rocio Sevilla	N/S
Pseudohipoparatiroidism	H. Cruces Baracaldo	10
	H. Conxo. Santiago	2
Retinitis pigmentosa	F. Jiménez Díaz	150
	H. V. Rocio Sevilla	N/S
Retinitis pigmentosa autosomic dominant (genes RHO y RDS)	F. Jiménez Díaz	25
	H. V. Rocio Sevilla	N/S
Retinitis pigmentosa X linked	F. Jiménez Díaz	50
	H. San Pablo Barcelona	6
	H. V. Rocio Sevilla	N/S
Retinitis pigmentosa, autosomal recessive	H. San Pablo Barcelona	135
	H. V. Rocio Sevilla	N/S
Retinoschisis, X-linked juvenile (RS1)	F. Jiménez Díaz	30

	H. La Fe. Valencia	29
Rheumathoid artritis (HLA markers)	Policlinica Gipuzkoa. San Sebastián	N/S
Rickets D-vit resistant VDR gene	H. Valle Hebrón	> 400
Russel Silver syndrome	H. del Mar – UPF Barcelona	3
Sex determination	Hospital Sant Joan de Déu, Barcelona	5
SHOX gene linked conditions	H. La Paz. Madrid	85
SRY Gene (sexual development alterations)	H. Miguel Servet. Zaragoza	54
	H. La Paz. Madrid	19
	H. Central de Asturias.	11
	F. Jiménez Díaz	10
	H Virgen del Camino	8
	Hospital Clínico Barcelona	5
	H. Cruces Baracaldo	5
	H. Gregorio Marañón Madrid	5
	Centro Oncológico de Galicia. A Coruña	4
	Centro de Patología Celular. Barcelona	3
	U. Salamanca	2
	H. V. Rocio Sevilla	N/S
	H. V. Rocio Sevilla	N/S
SRY gene / Amilogenina gene	Centro de Análisis Genéticos. Zaragoza	10
Uniparental disomy 14	H. La Fe. Valencia	12
	Hospital Clínico Barcelona	N/S
Uniparental disomy 15	Centro de Análisis Genéticos. Zaragoza	3
Uniparental disomy 7	Centro de Análisis Genéticos. Zaragoza	2
Usher syndrome	H. La Fe. Valencia	96
WAGR syndrome	H. La Fe. Valencia	12
Wilson disease (ATP7B)	Hospital Clínico Barcelona	20
	Clínica Universitaria Pamplona	12
	Instituto Biología Genética Molecular. Valladolid	8
	Centro Oncológico de Galicia. A Coruña	N/S
	H. La Paz. Madrid	N/S

* Centro de Patología Celular. Barcelona Data of 2000

MITOCONDRIAL DISEASES

	Centre	N. tests 2001
Mitochondrial disorders : Leigh, MERRF, MELAS, Pearson, Kerans-Sayre, depleciones, etc	H. 12 de Octubre	215
	Universidad Zaragoza. Dpto Biologia Molecular	200
	Hospital Valle Hebron. Barcelona	90
Mitochondrial myopathy, encephalopathy, lactic acidosis and strokelike episodes (MELAS)	Laboratorio de Análisis Dr. Echevarne Barcelona	8
	Hospital Clínico Barcelona	N/S
	Centro de Análisis Genéticos. Zaragoza	2
MERRF	Laboratorio de Análisis Dr. Echevarne Barcelona	4
MELAS y MERRF	IRO Barcelona	6

PHARMACOGENETICS

CYP2D6	Laboratorio de Análisis Dr. Echevarne Barcelona	41
CYP2C19	Laboratorio de Análisis Dr. Echevarne Barcelona	12
NAT2	Laboratorio de Análisis Dr. Echevarne Barcelona	15

Annex 6. Hereditary diseases tested and centres classified by autonomous regions

ANDALUCIA

Population: 7.403.968 inhabitants.

There are five public hospitals which perform genetic tests and one private laboratory that ave started activity very recently.

The number of hereditary diseases that can be tested in those centres is 64.

NEUROLOGIC CONDITIONS	Centres	N. test 2001 (N/S when not specified)
Alzheimer disease (Presenilina 1, APP, APOE)	H. Macarena Sevilla	20
	NBT DIAGEN Sevilla	N/S
Angelman syndrome	H. V. Rocio Sevilla	N/S
	H. Virgen de las Nieves. Granada	N/S
Dentatorubral-pallidoluysian atrophy (DRPLA)	H. Macarena Sevilla	N/S
	H. V. Rocio Sevilla	N/S
Dominant Ataxias (ADCAS) SCA1; SCA2 (Machado) SCA3; SCA6; SCA7; SCA8; SCA9;SCA10; SCA12	H. Macarena Sevilla	N/S (250 among all ataxias)
	H. V. Rocio Sevilla	N/S
Dominant macular dystrophies (RDS/periferina gene)	H. V. Rocio Sevilla	N/S
Duchenne - Becker muscular dystrophy	H. San Cecilio Granada	60
	H. V. Rocio Sevilla	N/S
	NBT DIAGEN Sevilla	N/S
Facioscapulohumeral muscular dystrophy 1A (FSHMD1A)	H. V. Rocio Sevilla	N/S
Fragile-X syndrome (FMR1)	H. Macarena Sevilla	225
	H. San Cecilio Granada	90
	H. V. Rocio Sevilla	N/S
	H. Virgen de las Nieves. Granada	N/S
Friedrich Ataxia	H. Macarena Sevilla	N/S
	H. V. Rocio Sevilla	N/S
	H. Virgen de las Nieves. Granada	N/S
Hirschsprung disease	H. V. Rocio Sevilla	N/S
Huntington disease	H. Macarena Sevilla	150
	H. San Cecilio Granada	25
	H. V. Rocio Sevilla	N/S
	H. Virgen de las Nieves. Granada	N/S
	NBT DIAGEN Sevilla	N/S
Kallmann syndrome	H. V. Rocio Sevilla	N/S
Kennedy disease (AR, exon 1)	H. Macarena Sevilla	6
	H. V. Rocio Sevilla	N/S
Leber optic atrophy	H. Macarena Sevilla	20
Miller-Dieker syndrome	H. V. Rocio Sevilla	N/S
Myotonic dystrophy Steinert	H. Macarena Sevilla	300
	H. V. Rocio Sevilla	N/S
	H. Virgen de las Nieves. Granada	N/S
	NBT DIAGEN Sevilla	N/S
Prader Willi syndrome	H. V. Rocio Sevilla	N/S
	H. Virgen de las Nieves. Granada	N/S
	NBT DIAGEN Sevilla	N/S
Rett syndrome	H. V. Rocio Sevilla	N/S
Smith-Magenis syndrome	H. V. Rocio Sevilla	N/S

Spinal muscular atrophy (Werdnig-Hoffmann y Kugelberg-Welander) (SMN)	H. Macarena Sevilla	N/S
	H. V. Rocio Sevilla	N/S
Williams-Beuren syndrome (LOH , FISH) (ATP7B)	H. V. Rocio Sevilla	N/S
	H. Virgen de las Nieves. Granada	N/S
Wolf-Hirshhorm syndrome	H. V. Rocio Sevilla	N/S
HEMATOLOGIC CONDITIONS		
ACE Angiotensin I convertor enzyme	NBT DIAGEN Sevilla	N/S
Alpha thalassemia	H. Virgen de las Nieves. Granada	N/S
Beta thalassemia	H. Virgen de las Nieves. Granada	N/S
Factor II G20210A	NBT DIAGEN Sevilla	N/S
Leukemias	H. Carlos Haya	147
	H. Virgen de las Nieves. Granada	N/S
	NBT DIAGEN Sevilla	N/S
Lymphomas	NBT DIAGEN Sevilla	N/S
Metilen TetraHidrofolato Reductasa (MTHFR)	NBT DIAGEN Sevilla	N/S
PAI-I Plasminogen activator inhibitor	NBT DIAGEN Sevilla	N/S
Thrombophilia (Mut. G1691A factor V) (Mut G20210A protrombina)	H. V. Rocio Sevilla	N/S
	H. Carlos Haya	130 (both)
METABOLIC CONDITIONS		
Alpha-1-antitripsina deficiency	H. Macarena Sevilla	50
	H. V. Rocio Sevilla	N/S
	NBT DIAGEN Sevilla	N/S
Apolipoprotein B (APOB) deficiency	NBT DIAGEN Sevilla	N/S
NEOPLASIC CONDITIONS		
Adenomatous polyposis of the colon (APC)	H. V. Rocio Sevilla	N/S
	H. Virgen de las Nieves. Granada	N/S
	NBT DIAGEN Sevilla	N/S
Breast cancer (BCRA1 y BCRA2)	H. V. Rocio Sevilla	N/S
Familiar medullar thyroid cancer FMTC	H. V. Rocio Sevilla	N/S
Familiar feochromocitome	H. V. Rocio Sevilla	N/S
	NBT DIAGEN Sevilla	N/S
Li-Fraumeni disease (p53)	H. V. Rocio Sevilla	N/S
	NBT DIAGEN Sevilla	N/S
Lynch syndrome	H. V. Rocio Sevilla	N/S
	H. Virgen de las Nieves. Granada	N/S
	NBT DIAGEN Sevilla	N/S
Multiple endocrine neoplasia, type IIA (MEN2A)	H. V. Rocio Sevilla	N/S
	H. Virgen de las Nieves. Granada	N/S
	NBT DIAGEN Sevilla	N/S
Multiple Endocrine Neoplasia type II B (MEN2B)	H. V. Rocio Sevilla	N/S
	NBT DIAGEN Sevilla	N/S
p53 EXONES 4,5,6,7,8 (Lung, breast, ovary, endometrio)	H. V. Rocio Sevilla	N/S
	H. Macarena Sevilla	N/S
Thyroid sporadic cancer	H. V. Rocio Sevilla	N/S
Von Hippel Lindau disease	H. V. Rocio Sevilla	N/S
OTHER CONDITIONS		
Absence of deferent conduct	NBT DIAGEN Sevilla	N/S
AIDS (□32-CCR5, V64I-CCR2, SDF1-3´A, RANTES-403C>A, RANTES-28C>G)	H. V. Rocio Sevilla	N/S
Azoospermia and oligospermia	H. Macarena Sevilla	50
	NBT DIAGEN Sevilla	N/S
Cavernomatosis	H. Macarena Sevilla	140
Chimerism determination for bone marrow transplant	H. Carlos Haya	221
Cystic fibrosis	H. Macarena Sevilla	180
	H. San Cecilio Granada	30
	H. Virgen de las Nieves. Granada	N/S

	H. V. Rocio Sevilla	N/S
	NBT DIAGEN Sevilla	N/S
- 68 -xxiiiDeafness, linked to Conexina 26 gene	H. V. Rocio Sevilla	N/S
Di George syndrome	H. V. Rocio Sevilla	N/S
Factor V Deficiency	H. Carlos Haya	67
	H. V. Rocio Sevilla	N/S
	H. Virgen de las Nieves. Granada	N/S
	NBT DIAGEN Sevilla	N/S
Hemochromatosis (HFE)	H. Macarena Sevilla	200
	H. San Cecilio Granada	60
	H. Virgen de las Nieves. Granada	N/S
	H. V. Rocio Sevilla	N/S
	NBT DIAGEN Sevilla	N/S
Homocystinuria	H. Virgen de las Nieves. Granada	N/S
Mediterranean familiar fever	H. V. Rocio Sevilla	N/S
Non Hodgking lynphomes: bcl2/IgH y Bcl1/IgH	H. Carlos Haya	68
Chimerism determination for bone marrow transplant	H. Virgen de las Nieves. Granada	N/S
Protrombine gene	H. Carlos Haya	63
	H. V. Rocio Sevilla	N/S
	H. Virgen de las Nieves. Granada	N/S
Retinitis pigmentosa	H. V. Rocio Sevilla	N/S
Retinitis pigmentosa autosomic dominant (genes RHO y RDS)	H. V. Rocio Sevilla	N/S
Retinitis pigmentosa, autosomal recessive	H. V. Rocio Sevilla	N/S
Retinoschisis, X-linked juvenile (RS1)	H. V. Rocio Sevilla	N/S
SRY gene	H. V. Rocio Sevilla	N/S

ARAGÓN

Population: 1.199.753 inhabitants.

There are 3 centres in which genetic tests are performed, of them 2 are public centres (1hospital and 1 university centre) and 1 private centre.

The number of conditions that can be tested in those centres is 55.

NEUROLOGIC CONDITIONS	Centres	N. test 2001
Alzheimer disease(Presenilina 1, APP, APOE)	H. Miguel Servet	23
	Centro de Análisis Genéticos. Zaragoza	7
Angelman syndrome	H. Miguel Servet	35
CADASIL	Centro de Análisis Genéticos. Zaragoza	1
Charcot-Marie-Tooth neuropathy	Centro de Análisis Genéticos. Zaragoza	22
Dominant ataxias (ADCAS) SCA1; SCA2 (Machado) SCA3; SCA6; SCA7; SCA8; SCA9; SCA10; SCA12	Centro de Análisis Genéticos. Zaragoza	13
Duchenne - Becker muscular dystrophy	Centro de Análisis Genéticos. Zaragoza	7
Fragile-X syndrome	H. Miguel Servet	129
	Centro de Análisis Genéticos. Zaragoza	73
Friedrich Ataxia (X25)	Centro de Análisis Genéticos. Zaragoza	8
Huntington disease	Centro de Análisis Genéticos. Zaragoza	9
	H. Miguel Servet	4
Kennedy disease. (AR, exón 1)	Centro de Análisis Genéticos. Zaragoza	1
Leber optic atrophy	Universidad Zaragoza. Dpto Biologia Molecular	20
	Centro de Análisis Genéticos. Zaragoza	3
MELAS	Centro de Análisis Genéticos. Zaragoza	2
	H. Miguel Servet	22
Myotonic dystrophy Steinert	Centro de Análisis Genéticos. Zaragoza	15
Neurofibromatosis type 1	Centro de Análisis Genéticos. Zaragoza	11
Neurophathy due to pressure sensitivity	Centro de Análisis Genéticos. Zaragoza	32
Norrie disease	Centro de Análisis Genéticos. Zaragoza	1
Prader Willi syndrome	H. Miguel Servet	6
	Centro de Análisis Genéticos. Zaragoza	2
Spinal muscular atrophy (Werdnig-Hoffmann y Kugelberg-Welander) (SMN)	Centro de Análisis Genéticos. Zaragoza	11
Torsion dystonia (DYT1)	Centro de Análisis Genéticos. Zaragoza	4
HEMATOLOGIC CONDITIONS		
a-Thalassemia	Centro de Análisis Genéticos. Zaragoza	11
ß-Thalassemia	Centro de Análisis Genéticos. Zaragoza	4
Thrombophilia (Mut G1691A factor V) (Mut G20210A protrombina)	Centro de Análisis Genéticos. Zaragoza	222/236
METABOLIC CONDITIONS		
Apolipoprotein B (APOB) deficiency	Universidad Zaragoza. Dpto Biologia Molecular	930
Gaucher disease	Universidad Zaragoza. Dpto Biologia Molecular	68
Hypercholesterolemia	Universidad Zaragoza. Dpto Biologia Molecular	350
Hyperliproteinemia type III, disbetalipoproteinemia (Hiperlipoproteinemia type III)	Universidad Zaragoza. Dpto Biologia Molecular	25
Mucolipidosis III	Centro de Análisis Genéticos. Zaragoza	1
Smitz-Lemli-Opitz síndrome 7- Esterol Reductasa (DRCR7)	Centro de Análisis Genéticos. Zaragoza	1

NEOPLASIC CONDITIONS		
Adenomatous polyposis of the colon (APC)	Centro de Análisis Genéticos. Zaragoza	1
Beta tubulina	H. Miguel Servet	10
Breast cancer (BCRA1 y BCRA2)	Centro de Análisis Genéticos. Zaragoza	2
Familiar feochromocitome	Centro de Análisis Genéticos. Zaragoza	1
Familiar melanoma (pl5, p19,CDK4)	Centro de Análisis Genéticos. Zaragoza	2
Li-Fraumeni disease (p53)	Centro de Análisis Genéticos. Zaragoza	2
Multiple Endocrine Neoplasia MEN2A	Centro de Análisis Genéticos. Zaragoza	2
Multiple Endocrine Neoplasia MEN2B	Centro de Análisis Genéticos. Zaragoza	2
p53 EXONES 4,5,6,7,8 (Lung, breast, ovary, endometrio)	H. Miguel Servet	105
	Centro de Análisis Genéticos. Zaragoza	2
Retinoblastoma	Centro de Análisis Genéticos. Zaragoza	1
Von Hippel Lindau disease	Centro de Análisis Genéticos. Zaragoza	2
MITOCONDRIAL DISEASES		
Mitochondrial disorders: Leigh, MERRF, MELAS, Pearson, Kerans-Sayre, etc	Universidad Zaragoza. Dpto Biologia Molecular	200
OTHER CONDITIONS		
Achondroplasia/Hipocondroplasia (F GFR3)	Centro de Análisis Genéticos. Zaragoza	2
	H. Miguel Servet	2
Azoospermia	H. Miguel Servet	36
	Centro de Análisis Genéticos. Zaragoza	13
Cystic fibrosis	Centro de Análisis Genéticos. Zaragoza	36
	H. Miguel Servet	19
Deafness, linked to Conexina 26 gene	Centro de Análisis Genéticos. Zaragoza	2
Deafness, rRNA 12S mitochondrial gene (Conexina 26)	Centro de Análisis Genéticos. Zaragoza	1
Di George syndrome	H. Miguel Servet	15 (FISH)
Factor V deficiency	H. Miguel Servet	1241
	Centro de Análisis Genéticos. Zaragoza	222
Hemochromatosis	H. Miguel Servet	410
	Centro de Análisis Genéticos. Zaragoza	312
Mediterranean familiar fever	Centro de Análisis Genéticos. Zaragoza	2
MTHFR	H. Miguel Servet	1241
Norrie disease(NDP)	Centro de Análisis Genéticos. Zaragoza	1
Polycystic Kidney Disease 1	Centro de Análisis Genéticos. Zaragoza	4
Protrombine gene	H. Miguel Servet	1241
	Centro de Análisis Genéticos. Zaragoza	236
SRY gene	H. Miguel Servet	54
SRY gene / gen Amilogenina	Centro de Análisis Genéticos. Zaragoza	10
Uniparental disomy 7	Centro de Análisis Genéticos. Zaragoza	2
Uniparental disomy 15	Centro de Análisis Genéticos. Zaragoza	3

ASTURIAS

Population: 1.075.329 inhabitants.

There is only 1 centre, a public hospital, in which genetic tests are performed

The number of conditions that can be tested in this centre is 25.

NEUROLOGIC CONDITIONS	Centres	N. test 2001
Alzheimer disease(Presenilina 1, APP, APOE)	H. Central de Asturias	220
Angelman syndrome	H. Central de Asturias	13
Charcot-Marie-Tooth neuropathy	H. Central de Asturias	6
Dentatorubral-pallidoluysian atrophy (DRPLA)	H. Central de Asturias	5
Dominant ataxias (ADCAS) SCA1; SCA2 (Machado) SCA3; SCA6; SCA7; SCA8; SCA9; SCA10; SCA12	H. Central de Asturias	25
Fragile-X syndrome	H. Central de Asturias	17
Friedrich ataxia	H. Central de Asturias	7
Huntington disease	H. Central de Asturias	15
Kennedy disease. (AR, exón 1)	H. Central de Asturias	5
Leber optic atrophy	H. Central de Asturias	2
Myotonic dystrophy Steinert	H. Central de Asturias	6
Parkinson (Parkin, a-sinucleina)	H. Central de Asturias	18
Prader Willi syndrome	H. Central de Asturias	13
Williams-Beuren syndrome (LOH , FISH) (ATP7B)	H. Central de Asturias	5
HEMATOLOGIC CONDITIONS		
Thrombophilia (Mut G1691A factor V) (Mut G20210A protrombina)	H. Central de Asturias.	127
METABOLIC CONDITIONS		
Adrenal hyperplasia, congenital	H. Central de Asturias.	17
OTHER CONDITIONS		
Achondroplasia/Hipocondroplasia (FGFR3)	H. Central de Asturias.	3
Azoospermia	H. Central de Asturias.	21
Cystic fibrosis	H. Central de Asturias.	9
Factor V deficiency	H. Central de Asturias.	127
Nefronoptisis (NPHP1)	H. Central de Asturias.	3
Polycystic Kidney Disease 1	H. Central de Asturias.	13
Polycystic Kidney Disease 2	H. Central de Asturias.	7
Protrombine gene	H. Central de Asturias.	127
SRY gene	H. Central de Asturias.	11

BALEARES

Population: 878.627 inhabitants.

There is only 1 centre, a public hospital, in which genetic tests are performed.

The number of conditions that can be tested in that centre is 19.

NEUROLOGIC CONDITIONS	Centres	N. test 2001
Amiloidean familiar polineuropathy	H. Son Dureta Mallorca	10
Angelman syndrome	H. Son Dureta Mallorca	25
Fragile-X syndrome	H. Son Dureta Mallorca	170
Huntington disease	H. Son Dureta Mallorca	13
Mental retardation FRAXE type linked (FMR2)	H. Son Dureta Mallorca	10
Miller-Dieker syndrome	H. Son Dureta Mallorca	6
Prader Willi syndrome	H. Son Dureta Mallorca	25
Silver-Russel syndrome	H. Son Dureta Mallorca	5
Smith-Magenis syndrome	H. Son Dureta Mallorca	10
Torsion dystonia (DYT1)	H. Son Dureta Mallorca	4
Williams-Beuren syndrome (LOH , FISH) (ATP7B)	H. Son Dureta Mallorca	15
HEMATOLOGIC CONDITIONS		
Chimerism determination for bone marrow transplant	H. Son Dureta Mallorca	60
OTHER CONDITIONS		
Achondroplasia/Hipocondroplasia (FGFR3)	H. Son Dureta Mallorca	5
Azoospermia	H. Son Dureta Mallorca	50
Cystic fibrosis	H. Son Dureta Mallorca	110
Di George syndrome	H. Son Dureta Mallorca	35
Hemochromatosis	H. Son Dureta Mallorca	20
Polycystic Kidney Disease 1	H. Son Dureta Mallorca	N/S
Polycystic Kidney Disease 2	H. Son Dureta Mallorca	N/S

CANARIAS

Population: 1.781.366 inhabitants.

There are 2 centres in which genetic tests are performed, both of them public hospitals.

The number of conditions that can be tested in those centres is 21.

NEUROLOGIC CONDITIONS	Centres	N. test 2001
Alzheimer disease(Presenilina 1, APP, APOE)	H. Universitario Tenerife	20
Angelman syndrome	H.Universitario Las Palmas	1
Duchenne - Becker muscular dystrophy	H.Universitario Las Palmas	26
Fragile-X syndrome	H.Universitario Las Palmas	93
Mental retardation FRAXE type linked (FMR2)	H.Universitario Las Palmas	93
Myotonic dystrophy Steinert	H.Universitario Las Palmas	19
Prader Willi syndrome	H.Universitario Las Palmas	34
Williams-Beuren syndrome (LOH , FISH) (ATP7B)	H.Universitario Las Palmas	14
	H.Universitario Las Palmas	9
HEMATOLOGIC CONDITIONS		
Thrombophilia (Mut G1691A factor V) (Mut G20210A protrombina)	H. Universitario Tenerife	50
METABOLIC CONDITIONS		
Alpha-1-antitripsina deficiency	H.Universitario Las Palmas	5
Primary hyperaldosteronism	H. Universitario Tenerife	2
Primary hyperoxaluria	H. Universitario Tenerife	20
OTHER CONDITIONS		
Achondroplasia/Hipocondroplasia (FGFR3)	H.Universitario Las Palmas	2
Azoospermia	H. Universitario Tenerife	24
Cystic fibrosis	H.Universitario Las Palmas	43
	H. Universitario Tenerife	18
Deafness, linked to Conexina 26 gene	H.Universitario Las Palmas	31
Factor V deficiency	H. Universitario Tenerife	20
Hemochromatosis	H. Universitario Tenerife	64
Nefronoptisis (NPHP1)	H. Universitario Tenerife	2
Protrombine gene	H. Universitario Tenerife	20

CANTABRIA

Population: 537.606 inhabitants.

There is only 1 centre, a public hospital, in which genetic tests are performed. The number of conditions that can be tested in that centre is 21.

NEUROLOGIC CONDITIONS	Centres	N. test 2001
Alzheimer disease(Presenilina 1, APP, APOE)	H. Valdecilla	25
Charcot-Marie-Tooth neuropathy	H. Valdecilla	25
Friedrich ataxia	H. Valdecilla	20
Myotonic dystrophy Steinert	H. Valdecilla	20
Neurophathy due to pressure sensitivity	H. Valdecilla	25
Schizophrenia	H. Valdecilla	102
Tuberose sclerosis (Gen TSC1,Gen TSC2)	H. Valdecilla	N/S
HEMATOLOGIC CONDITIONS		
Thrombophilia (Mut. G1691A factor V) (Mut. G20210A protrombina)	H. Valdecilla	14
Thrombophilia (Mut G20210A, protrombina)	H. Valdecilla	14
Thrombophilia (Mut C677T)	H. Valdecilla	14
Thrombophilia (metilentetrahidrofolatoreductasa)	H. Valdecilla	14
INMUNODEFICIENCIES		
B. Cesar-Chediak-Higashi disease	H. Valdecilla	N/S
Complement deficiencies	H. Valdecilla	N/S
Di George syndrome	H. Valdecilla	4
HLA type II molecules deficiency	H. Valdecilla	N/S
Inmunologic poligenetic diseases screening	H. Valdecilla	>200 (each)
LAD deficiencies type I and type II	H. Valdecilla	N/S
Omenn syndrome Mut gene RAG1 Mut gene RAG2	H. Valdecilla	1 1
Wisckott-Aldrich syndrome	H. Valdecilla	N/S
NEOPLASIC CONDITIONS		
Adenomatous polyposis of the colon (APC) Gen APC	H. Valdecilla	5
Adenomatous polyposis of the colon (APC) Gen P53, Gen kRAS, Gen hMHS2, Gen hMLH1, Gen hPMS1,Gen hPMS2		5 (each mutation)
OTHER CONDITIONS		
Mediterranean familiar fever Mut. M694V Mut V726A	H. Valdecilla	 17 17

CASTILLA-LA MANCHA

Population: 1.755.053 inhabitants.
There is only 1 centre, a public hospital, in which genetic tests are performed. The number of conditions that can be tested in that centre is 6.

NEUROLOGIC CONDITIONS	Centres	N. test 2001
Angelman syndrome	H. Virgen de la Salud Toledo	4
Fragile-X syndrome	H. Virgen de la Salud Toledo	63
Prader Willi syndrome	H. Virgen de la Salud Toledo	6
Williams-Beuren syndrome (LOH , FISH) (ATP7B)	H. Virgen de la Salud Toledo	5
HEMATOLOGIC CONDITIONS		
Leukemia and limphoma	H. Virgen de la Salud Toledo	375
OTHER CONDITIONS		
Azoospermia	H. Virgen de la Salud Toledo	86

CASTILLA - LEÓN

Population: 2.479.425 inhabitants.
There are 2 centres in which genetic tests are performed, both of them are public university centres. The number of conditions that can be tested in those centres is 22.

NEUROLOGIC CONDITIONS	Centres	N. test 2001
Alzheimer disease(Presenilina 1, APP, APOE)	U. Salamanca	50
Friedrich ataxia	Instituto Biología Genética Molecular. Valladolid	4
Fragile-X syndrome	Instituto Biología Genética Molecular. Valladolid	53
Kennedy disease. (AR, exón 1)	Instituto Biología Genética Molecular. Valladolid	1
Parkinson (Parkin, a-sinucleina)	U. Salamanca	12
Spinal muscular atrophy (Werdnig-Hoffmann y Kugelberg-Welander) (SMN)	Instituto Biología Genética Molecular. Valladolid	2
NEOPLASIC CONDITIONS		
Breast cancer (BCRA1 y BCRA2)	U. Salamanca	50
CK-19	U. Salamanca	12
GST mu (colon, vejiga)	U. Salamanca	400
K-RAS (CODON 12, 13) (lung, colon, ovary, larynx)	U. Salamanca	25
Li-Fraumeni disease (p53)	U. Salamanca	10
p53 EXONES 4,5,6,7,8 (Lung, breast, ovary, endometrio)	U. Salamanca	200
Von Hippel Lindau disease	U. Salamanca	5
METABOLIC CONDITIONS		
Alpha-1-antitripsina deficiency	Instituto Biología Genética Molecular. Valladolid	15
OTHER CONDITIONS		
Azoospermia	U. Salamanca	25
Celiac disease	Instituto Biología Genética Molecular. Valladolid	150
Cystic fibrosis	Instituto Biología Genética Molecular. Valladolid	200
Factor V deficiency	U. Salamanca	50
Hemochromatosis	U. Salamanca Instituto Biología Genética Molecular. Valladolid	75 60
Protrombine gene	U. Salamanca	50
SRY gene	U. Salamanca	2
Wilson disease (ATP7B)	Instituto Biología Genética Molecular. Valladolid	8

CATALUÑA

Population: 6.361.365 inhabitants.

There are 9 centres in which genetic tests are performed, of them 5 are public hospitals centres and 4 private centres.

The number of conditions that can be tested in those centres is 70.

NEUROLOGIC CONDITIONS	Centres	N. test 2001
Alzheimer disease(Presenilina 1, APP, APOE)	Hospital Clínico Barcelona	300
	Laboratorio de Análisis Dr. Echevarne Barcelona	9
Aneuploidies due toQF-PCR	Laboratorio de Análisis Dr. Echevarne Barcelona	460
Angelman syndrome	Hospital Sant Joan de Déu, Barcelona	44
	H. Parc Tauli Barcelona	23
	H. del Mar – UPF Barcelona	2
	Laboratorio de Análisis Dr. Echevarne Barcelona	2
Charcot-Marie-Tooth neuropathy	IRO Barcelona	313
	Hospital Sant Joan de Déu, Barcelona	71
	Laboratorio de Análisis Dr. Echevarne Barcelona	14
Dentatorubral-pallidoluysian atrophy (DRPLA) (Dentatorubropalidoluisiana)	IRO Barcelona	1
	Laboratorio de Análisis Dr. Echevarne Barcelona	2
Dominant ataxias (ADCAS) SCA1; SCA2 (Machado) SCA3; SCA6; SCA7; SCA8; SCA9; SCA10; SCA12	IRO Barcelona	203
	Laboratorio de Análisis Dr. Echevarne Barcelona	64
Duchenne - Becker muscular dystrophy	H. San Pablo Barcelona	163
	Hospital Sant Joan de Déu, Barcelona	14
	Laboratorio de Análisis Dr. Echevarne Barcelona	7
Fragile-X syndrome	Hospital Sant Joan de Déu, Barcelona	308
	Laboratorio de Análisis Dr. Echevarne Barcelona	39
	H. del Mar – UPF Barcelona	10
Friedrich ataxia	Hospital Sant Joan de Déu, Barcelona	25
	IRO Barcelona	9
	Laboratorio de Análisis Dr. Echevarne Barcelona	8
	Hospital Clínico Barcelona	N/S
Fronto-temporal dementia	Hospital Clínico Barcelona	30
Huntington disease	Laboratorio de Análisis Dr. Echevarne Barcelona Hospital Clínico Barcelona	20
Kennedy disease. (AR, exón 1)	H. San Pablo Barcelona	6
	Laboratorio de Análisis Dr. Echevarne Barcelona	4
Leber optic atrophy	IRO Barcelona	19
	Laboratorio de Análisis Dr. Echevarne Barcelona	5
Mental retardation FRAXE type	Hospital Clínico Barcelona	
MELAS	Laboratorio de Análisis Dr. Echevarne Barcelona	8
MELAS y MERRF	IRO Barcelona	6
MERRF	Laboratorio de Análisis Dr. Echevarne Barcelona	4
Myotonic distrophy of waists (LGMD)	H. San Pablo Barcelona	25
Myotonic dystrophy Steinert	H. San Pablo Barcelona	263
	Hospital Sant Joan de Déu, Barcelona	14

	Laboratorio de Análisis Dr. Echevarne Barcelona	12
NARP	Laboratorio de Análisis Dr. Echevarne Barcelona	2
Neurofibromatosis type 1	IRO Barcelona	356
	Laboratorio de Análisis Dr. Echevarne Barcelona	3
	Hospital Sant Joan de Déu, Barcelona	15
Neurofibromatosis type 2	IRO Barcelona	13 NF2
Neurophathy due to pressure sensitivity	Hospital Sant Joan de Déu, Barcelona	23
	Laboratorio de Análisis Dr. Echevarne Barcelona	3
Parkinson (Parkin, a-sinucleina)	Hospital Clínico Barcelona	25
Prader Willi syndrome	Hospital Sant Joan de Déu, Barcelona	74
	H. Parc Tauli Barcelona	30
	Laboratorio de Análisis Dr. Echevarne Barcelona	7
	H. del Mar – UPF Barcelona	3
Rett syndrome	Hospital Sant Joan de Déu, Barcelona	244
Smith-Magenis syndrome	H. del Mar – UPF Barcelona	3
Spinal muscular atrophy (Werdnig-Hoffmann y Kugelberg-Welander) (SMN)	H. San Pablo Barcelona	298
	Laboratorio de Análisis Dr. Echevarne Barcelona	8
Williams-Beuren syndrome (LOH , FISH) (ATP7B)	H. del Mar – UPF Barcelona	30
	H. Parc Tauli Barcelona	5
	Hospital Sant Joan de Déu, Barcelona	26
Wolfram syndrome	IRO Barcelona	8
HEMATOLOGIC CONDITIONS		
Alpha Thalassemia	H. San Pablo Barcelona	20
	Laboratorio de Análisis Dr. Echevarne Barcelona	9
Beta Thalassemia	H. San Pablo Barcelona	46
	Laboratorio de Análisis Dr. Echevarne Barcelona	3
Drepanocitosis	H. San Pablo Barcelona	5
Haemophilia A	H. San Pablo Barcelona	113
	Laboratorio de Análisis Dr. Echevarne Barcelona	3
Haemophilia B	H. San Pablo Barcelona	22
	Laboratorio de Análisis Dr. Echevarne Barcelona	1
Leukemias	H. del Mar – UPF Barcelona	800
	Laboratorio de Análisis Dr. Echevarne Barcelona	16
Thrombophilia (Mut G1691A factor V) (Mut G20210A protrombina)	Laboratorio de Análisis Dr. Echevarne Barcelona	252
	IRO Barcelona	85
NEOPLASIC CONDITIONS		
Adenomatous polyposis of the colon (APC)	Laboratorio de Análisis Dr. Echevarne Barcelona	7
Breast cancer (BCRA1 y BCRA2)	H. San Pablo Barcelona	79
	H. del Mar – UPF Barcelona	20
	Laboratorio de Análisis Dr. Echevarne Barcelona	2
Familiar medullar thyroid cancer FMTC	Hospital Clínico Barcelona	N/S
Li-Fraumeni disease (p53)	H. San Pablo Barcelona	8
	Laboratorio de Análisis Dr. Echevarne Barcelona	3
Multiple Endocrine Neoplasia type 1 (MEN1)	Hospital Clínico Barcelona	N/S
Non polyposic colonic cancer	Hospital Clínico Barcelona	N/S
p53 EXONES 4,5,6,7,8 (Lung, breast, ovary, endometrio)	Laboratorio de Análisis Dr. Echevarne Barcelona	5
Von Hippel-Lindau	Hospital Clínico Barcelona	N/S
METABOLIC CONDITIONS		
5 a-reductasa deficiency type 2 Gene SRD5A2	H. Valle Hebrón Barcelona	5

17B-hidroxiesteroide-deshidrogenasa deficiency Gene 17□-HSD3	H. Valle Hebrón	2
21-hidroxilasa deficiency	Hospital Sant Joan de Déu, Barcelona	51
	Laboratorio de Análisis Dr. Echevarne Barcelona	3
Alpha-1-antitripsina deficiency	Laboratorio de Análisis Dr. Echevarne Barcelona	16
	Hospital Clínico Barcelona	15
Maturity-onset diabetes of the young MODY type (GK, HNF1)	Hospital Clínico Barcelona	N/S
GH deficiency. Gene GH1	H. Valle Hebrón Barcelona	> 100
Hunter disease	Hospital Clínico Barcelona	N/S
Hurler disease	Laboratorio de Análisis Dr. Echevarne Barcelona	1
	Hospital Clínico Barcelona	N/S
Smitz-Lemli-Opitz syndrome 7- Esterol Reductasa (DRCR7)	Hospital Clínico Barcelona	N/S
OTHER CONDITIONS		
Androgenic resistances: Morris syndrome, Reifenstein syndrome (AR exones 2-8) Gen AR	H. Valle Hebrón Barcelona	30
	Hospital Clínico Barcelona	N/S
Azoospermia	Hospital Clínico Barcelona	290
	Laboratorio de Análisis Dr. Echevarne Barcelona	39
	Centro de Patología Celular. Barcelona	61
Cistinuria	IRO Barcelona	20
Celiac disease	Laboratorio de Análisis Dr. Echevarne Barcelona	21
Cystic fibrosis	IRO Barcelona	574
	Centro de Patología Celular. Barcelona	44
	Hospital Sant Joan de Déu, Barcelona	31
	Laboratorio de Análisis Dr. Echevarne Barcelona	13
	Hospital Clínico Barcelona	N/S
Di George syndrome	Centro de Patología Celular. Barcelona	9
Di George syndrome/ S. Velocardiofacial (FISH)	H. Parc Tauli Barcelona	32
Factor V deficiency	Laboratorio de Análisis Dr. Echevarne Barcelona	134
	IRO Barcelona	58
Gilbert syndrome	H. San Pablo Barcelona	190
Hemochromatosis	H. San Pablo Barcelona	385
	Hospital Clínico Barcelona	300
	Laboratorio de Análisis Dr. Echevarne Barcelona	257
Homocistinuria	IRO Barcelona	1
	Laboratorio de Análisis Dr. Echevarne Barcelona	31
Homocistinuria MTHFR-ts	Hospital Sant Joan de Déu, Barcelona	81
HPV	Laboratorio de Análisis Dr. Echevarne Barcelona	295
Mitochondrial disorders	Hospital Valle Hebron. Barcelona	90
Norrie disease (NDP)	Hospital Clínico Barcelona	N/S
	IRO Barcelona	52
	Laboratorio de Análisis Dr. Echevarne Barcelona	114
Rickets D-vit resistant VDR gene	H. Valle Hebrón Barcelona	> 400
Retinitis pigmentosa, autosomal recesive	H. San Pablo Barcelona	135
Retinitis pigmentosa, X-linked	H. San Pablo Barcelona	6
Russel Silver syndrome, primordial dwarf (Disomía uniparental cr. 7, duplicación 7p)	H. del Mar – UPF Barcelona	3
Sex determination	Hospital Sant Joan de Déu, Barcelona	5
SRY gene	Centro de Patología Celular. Barcelona	3
Wilson disease (ATP7B)	Hospital Clínico Barcelona	20

COMUNIDAD VALENCIANA

Population: 4.202.608 inhabitants.

There are 5 centres in which genetic tests are performed, of them 3 are public centres (1 hospital and 2 university centres) and 2 private.

The number of conditions that can be tested in those centres is 57

NEUROLOGIC CONDITIONS	Centres	N. test 2001
Alzheimer disease(Presenilina 1, APP, APOE)	ANCOR Alicante	N/S
Angelman syndrome	H. La Fe. Valencia	35
	IGEM Valencia	2
Charcot-Marie-Tooth neuropathy	H. La Fe. Valencia	53
	IGEM Valencia	20
Dentatorubral-pallidoluysian atrophy (DRPLA) (Dentatorubropalidoluisiana)	H. La Fe. Valencia	37
Distrofia facio-escapulo-humeral	H. La Fe. Valencia	25
	IGEM Valencia	9
Dominant ataxias (ADCAS) SCA1; SCA2 (E. de Machado) SCA3; SCA6; SCA7; SCA8; SCA9 SCA10; SCA12	H. La Fe. Valencia	222
	IGEM Valencia	10
Duchenne - Becker muscular dystrophy	H. La Fe. Valencia	26
	IGEM Valencia	5
Fragile-X syndrome	H. La Fe. Valencia	295
	IGEM Valencia	50
	ANCOR Alicante	N/S
Friedrich ataxia	H. La Fe. Valencia	19
	IGEM Valencia	5
Huntington disease	H. La Fe. Valencia	65
	IGEM Valencia	3
Kennedy disease. (AR, exón 1)	H. La Fe. Valencia	5
Mental retardation FRAXE type linked (FMR2)	H. La Fe. Valencia	100
Myotonic dystrophy Steinert	H. La Fe. Valencia	66
	IGEM Valencia	4
Neurophathy due to pressure sensitivity	H. La Fe. Valencia	9
	IGEM Valencia	5
Prader Willi syndrome	H. La Fe. Valencia	60
	IGEM Valencia	4
Spinal muscular atrophy (Werdnig-Hoffmann y Kugelberg-Welander) (SMN)	H. La Fe. Valencia	14
	IGEM Valencia	4
Torsion dystonia (DYT1)	H. La Fe. Valencia	4
	IGEM Valencia	1
Williams-Beuren syndrome (LOH , FISH) (ATP7B)	H. La Fe. Valencia	(loh) 24
	IGEM Valencia	2
HEMATOLOGIC CONDITIONS		
Haemophilia A	IGEM Valencia	2
	ANCOR Alicante	N/S
Haemophilia B	ANCOR Alicante	N/S
Inmunodeficiencia type Bruton	H. La Fe. Valencia	14
Leukemias	ANCOR Alicante	N/S
Thrombophilia (Mut G1691A factor V) (Mut G20210A protrombina)	ANCOR Alicante	N/S

METABÓLIC CONDITIONS		
Hunter disease	H. La Fe. Valencia	2
NEOPLASIC CONDITIONS		
Adenomatous polyposis of the colon (APC)	H. La Fe. Valencia	20
Breast cancer (BCRA1 y BCRA2)	ANCOR Alicante	N/S
Li-Fraumeni disease (p53)	ANCOR Alicante	N/S
Familiar feochromocitome	H. La Fe. Valencia	N/S
Instability in microsatelites MSI Panel 5 Markers	ANCOR Alicante	N/S
K-RAS (CODON 12, 13)(lung, colon, ovary, larynx)	ANCOR Alicante	N/S
Multiple Endocrine Neoplasia MEN2A	H. La Fe. Valencia	16
Multiple Endocrine Neoplasia MEN2B	H. La Fe. Valencia	N/S
p53 EXONES 4,5,6,7,8 (Lung, breast, ovary, endometrio) Sarcomas	U. Valencia	~50
	ANCOR Alicante	N/S
Retinoblastoma	H. La Fe. Valencia	26
Sporadic Cancer: kidney	U. Valencia	~60
Sporadic Cancer : bladder	U. Valencia	~40
Sporadic Cancer: Meningiomas	U. Valencia	~20
Sporadic Cancer: Neuroblastomas	U. Valencia	~70
Sporadic Cancer: Sarcomas	U. Valencia	~50
Von Hippel Lindau disease	H. La Fe. Valencia	8
OTHER CONDITIONS		
Beckwith-Wiedemann syndrome	H. La Fe. Valencia	6
Celiac disease	IGEM Valencia	4
Cystic fibrosis	H. La Fe. Valencia	179
	IGEM Valencia	5
Di George syndrome	H. La Fe. Valencia	84
	IGEM Valencia	6
Emery-Dreifuss disease	H. La Fe. Valencia	3
Epidermolisis bullosa distrofica	H. La Fe. Valencia	1
Factor V deficiency	ANCOR Alicante	N/S
Hemochromatosis	H. La Fe. Valencia	314
	U. Valencia	~100
	IGEM Valencia	5
	ANCOR Alicante	N/S
Hipercolesterolemia Familiar y gen SRY	U. Alicante	N/S
Incontinentia pigmenti	H. La Fe. Valencia	N/S
Mental retardation familiar	H. La Fe. Valencia	21
Miller-Diecker syndrome	H. La Fe. Valencia	3
Retinosquisis	H. La Fe. Valencia	29
Uniparental Disomy 14	H. La Fe. Valencia	12
Usher syndrome	H. La Fe. Valencia	96
WAGR syndrome	H. La Fe. Valencia	12

EXTREMADURA

Population: 1.073.381 inhabitants.

There is 1 centre, a public hospital, in which genetic tests are performed.

The number of conditions that can be tested in that centre is 4.

NEUROLOGIC CONDITIONS	Centres	N. test 2001
Angelman syndrome	H. Materno Infantil Badajoz	20
Prader Willi syndrome	H. Materno Infantil Badajoz	40
Smith-Magenis syndrome	H. Materno Infantil Badajoz	8
Williams-Beuren syndrome (LOH , FISH) (ATP7B)	H. Materno Infantil Badajoz	15

GALICIA

Population: 2.732.926 inhabitants.

There are 4 centres in which genetic tests are performed, 3 of them are public hospitals and 1 private.

The number of conditions that can be tested in those centres is 69.

NEUROLOGIC CONDITIONS	Centres	N. test 2001
Alzheimer disease(Presenilina 1, APP, APOE)	H. Conxo. Santiago	3
Angelman syndrome	Centro Oncológico de Galicia	4
	Hospital Clínico Santiago	8
Charcot-Marie-Tooth neuropathy	H. Conxo. Santiago	21
Dentatorubral-pallidoluysian atrophy (DRPLA) (Dentatorubropalidoluisiana)	Hospital Clínico Santiago	5
Dominant ataxias (ADCAS) SCA1; SCA2 (Machado) SCA3; SCA6; SCA7; SCA8; SCA9; SCA10; SCA12	H. Conxo. Santiago	29
	Hospital Clínico Santiago	104
Duchenne - Becker muscular dystrophy	Centro Oncológico de Galicia	4
	Hospital Clínico Santiago	6
Fragile-X syndrome	Centro Oncológico de Galicia	80
	Hospital Clínico Santiago	35
Friedrich ataxia	Hospital Clínico Santiago	20
	H. Conxo. Santiago	7
Huntington disease	H. Conxo. Santiago	29
	Hospital Clínico Santiago	25
Leber optic atrophy	H. Conxo. Santiago	2
Mental retardation FRAXE type linked (FMR2)	Hospital Clínico Santiago	50
Myotonic dystrophySteinert	Hospital Clínico Santiago	10
Neurophathy due to pressure sensitivity	H. Conxo. Santiago	11
Prader Willi syndrome	Centro Oncológico de Galicia	5
	Hospital Clínico Santiago	10
Rett syndrome	H. Conxo. Santiago	3
Smith-Magenis syndrome	H. Conxo. Santiago	1
	Hospital Clínico Santiago	5
Williams-Beuren syndrome (LOH , FISH) (ATP7B)	H. Conxo. Santiago	2
	Hospital Clínico Santiago	8
HEMATOLOGIC CONDITIONS		
Chimerism for bone marrow transplants	Centro Oncológico de Galicia	6
Leukemias	H. Conxo. Santiago	168
Leukemias y linfomas (traslocaciones)	Centro Oncológico de Galicia	225
Mut C>T 677 gene MTHFR	H. Conxo. Santiago	50
Thrombophilia (Mut G1691A factor V) (Mut G20210A protrombina)	Hospital Juan Canalejo	2160
	H. Conxo. Santiago	261
von Willebrand disease	Hospital Juan Canalejo	400
NEOPLASIC CONDITIONS		
Adenomatous polyposis of the colon (APC)	H. Conxo. Santiago	19
Breast cancer (BCRA1 y BCRA2)	H. Conxo. Santiago	13
Li-Fraumeni disease (p53)	Centro Oncológico de Galicia	N/S
	H. Conxo. Santiago	2
Multiple Endocrine Neoplasia MEN2A	H. Conxo. Santiago	4
Multiple Endocrine Neoplasia MEN2B	H. Conxo. Santiago	N/S
p53 EXONES 4,5,6,7,8 (Lung, breast, ovary, endometrio)	H. Conxo. Santiago	47
Tirosinasa (cel. Circulantes, cáncer de tiroides)	H. Conxo. Santiago	43
Tirosinasa (cel. Circulantes, melanoma)	H. Conxo. Santiago	16
Von Hippel Lindau disease	H. Conxo. Santiago	1

METABOLIC CONDITIONS		
3-beta-hidroxiesteroide-deshidrogenasa deficiency	H. Conxo. Santiago	1
11-beta-hidroxilasa deficiency	H. Conxo. Santiago	4
17-alfa-hidroxilasa deficiency	H. Conxo. Santiago	2
21-hidroxilasa deficiency	H. Conxo. Santiago	60
Adrenal hyperplasia, congenital	H. Conxo. Santiago	64
Alpha-1-antitripsina deficiency	H. Conxo. Santiago	6
Apolipoprotein B (APOB) deficiency	H. Conxo. Santiago	2
Cerebrotendinosis xantomatosa (CYP27)	H. Conxo. Santiago	1
Growth Hormone deficiency	H. Conxo. Santiago	43
Hypercholesterolemia	H. Conxo. Santiago	2
Insensibility to GH syndrome	H. Conxo. Santiago	71
Lipodistrofia Parcial Familiar	H. Conxo. Santiago	1
Primary hyperaldosteronism	H. Conxo. Santiago	5
Triple A syndrome	H. Conxo. Santiago	3
OTHER CONDITIONS		
Achondroplasia/Hipocondroplasia (FGFR3)	H. Conxo. Santiago	3
Alport síndrome	H. Conxo. Santiago	2
Androgenic Resistances: Morris syndrome, Reifenstein syndrome (AR exones 2-8)	H. Conxo. Santiago	3
Aneuploidias	Centro Oncológico de Galicia	80
Azoospermia	Centro Oncológico de Galicia Hospital Clínico Santiago	10 3
Cystic fibrosis	Centro Oncológico de Galicia H. Conxo. Santiago Hospital Clínico Santiago	60 5 23
Di George syndrome	Centro Oncológico de Galicia H. Conxo. Santiago Hospital Clínico Santiago	30 6 40
Factor V deficiency	H. Conxo. Santiago	43
Hemochromatosis	Centro Oncológico de Galicia H. Conxo. Santiago Hospital Clínico Santiago	30 187 10
Hipoplasia adrenal congénita	H. Conxo. Santiago	1
Holoprosencefalia	H. Conxo. Santiago	1
Homocistinuria	H. Conxo. Santiago	1
Mediterranean familiar fever	Hospital Clínico Santiago	2
NPC1 linkage	H. Conxo. Santiago	N/S
Protrombine gene	H. Conxo. Santiago	218
Panhipopituitarismo	H. Conxo. Santiago	11
Poliendrocrinopathy autoinmune type 1	H. Conxo. Santiago	1
Polycystic Kidney Disease 1	H. Conxo. Santiago	14
Polycystic Kidney Disease 2	H. Conxo. Santiago	2
Pseudohipoparatiroidismo	H. Conxo. Santiago	2
SRY gene	Centro Oncológico de Galicia	4
Wilson disease (ATP7B)	Centro Oncológico de Galicia	N/S

MADRID

Population: 5.372.433 inhabitants.

There are 9 centres in which genetic tests are performed, of them 8 are public centres (6 hospitals and 1university centres and 2 research centres) and 1 private hospital.

The number of conditions that can be tested in those centres is 57.

NEUROLOGIC CONDITIONS	Centres	N. test 2001
Angelman syndrome	F. Jiménez Díaz	25
Charcot-Marie-Tooth neuropathy	F. Jiménez Díaz	40
Criptic chomosomic alterations (FISH, Multitest-T y C)	Centro Investigación Anomalías Congénitas Madrid.	N/S
Dentatorubral-pallidoluysian atrophy (DRPLA)	F. Jiménez Díaz	135
Distrofias maculares dominantes (gen RDS/periferina)	F. Jiménez Díaz	50
Dominant ataxias (ADCAS) SCA1; SCA2.(Machado) SCA3; SCA6; SCA7; SCA8; SCA9, SCA10; SCA12	F. Jiménez Díaz	135
Duchenne - Becker muscular dystrophy	F. Jiménez Díaz H. La Paz. Madrid	45 20
Fragile-X syndrome	H. Ramón y Cajal F. Jiménez Díaz	195 110
Friedrich ataxia	F. Jiménez Díaz	135
Huntington disease	F. Jiménez Díaz	85
Kallman (microdel Xp22)	F. Jiménez Díaz	6
Myotonic dystrophySteinert	H. La Paz. Madrid F. Jiménez Díaz	80 80
Mental retardation FRAXE type linked (FMR2)	F. Jiménez Díaz	2
Neurofibromatosis	H. Ramón y Cajal	45
Neurophathy due to pressure sensitivity	F. Jiménez Díaz	40
Norrie disease	F. Jiménez Díaz	8
Prader Willi syndrome	F. Jiménez Díaz	25
Smith-Magenis syndrome	F. Jiménez Díaz	N/S
Spinal muscular atrophy (Werdnig-Hoffmann y Kugelberg-Welander) (SMN)	H. Ramón y Cajal	127
Torsion dystonia (DYT1)	F. Jiménez Díaz	20
Williams-Beuren syndrome (LOH , FISH) (ATP7B)	F. Jiménez Díaz	5
HEMATOLOGIC CONDITIONS		
Thrombophilia (Mut G1691A factor V) (Mut G20210A protrombina)	F. Jiménez Díaz H. Gregorio Marañón Madrid	35 N/S
NEOPLASIC CONDITIONS		
Adenomatous polyposis of the colon (APC)	CNIO Madrid H. San Carlos Madrid	10 10
Breast cancer (BCRA1 y BCRA2)	H. San Carlos Madrid CNIO Madrid	50 families 45
Familiar feochromocitome	CNIO Madrid	5
Familiar melanoma (pl6)	CNIO Madrid	5
HLXB9	CNIO Madrid	2
Li-Fraumeni disease	H. San Carlos Madrid	N/S
MEN1	CNIO Madrid	20
Multiple Endocrine Neoplasia MEN2A	CNIO Madrid	40
Non poliposic Colonic Cancer	CNIO Madrid	15
Other oncohematologic conditions	CNIO Madrid	300
p53 EXONES 4,5,6,7,8 (Lung, breast, ovary, endometrio)	H. San Carlos Madrid CNIO Madrid	130 10
PTEN	CNIO Madrid	5
Von Hippel Lindau disease	CNIO Madrid	30

METABOLIC CONDITIONS		
11-beta-hidroxilasa deficiency	H. Gregorio Marañón	4
21-hidroxilasa deficiency	H. Gregorio Marañón Madrid	400
	H. La Paz. Madrid	352
Adrenal hyperplasia, congenital	H. La Paz. Madrid	352
Alpha-1-antitripsina deficiency	H. Gregorio Marañón	N/S
	H. La Paz. Madrid	54
Carnitina palmitoil transferasa II deficiency, fenotype muscular, mutacións S113L	H. 12 de Octubre	148
Carnitina Palmitoil Transferasa II deficiency (Mutación Y628S)	H. 12 de Octubre	20
	Centro de Diagnóstico de Enfermedades Moleculares Madrid	1
Diabetes type MODY. (GK, HNF1)	H. La Paz. Madrid	76
Fenilcetonuria-Fenialaninemia (PAH)	Centro de Diagnóstico de Enfermedades Moleculares Madrid	45
GH deficiency	H. La Paz. Madrid	8
HLA of susceptibility to diabetes type I	H. La Paz. Madrid	24
Insensibility to GH	H. La Paz. Madrid	N/S
McArdle disease (défict de miofosforilasa). Mutaciones R49X, G204S y W797R	H. 12 de Octubre	134
Metil crotonil glicinuria (Genes MCCA y MCCB)	Centro de Diagnóstico de Enfermedades Moleculares Madrid	16
MCAD Medium Chain Acil-CoA deshidrogenasa deficiency (Mutation K304E)	Centro de Diagnóstico de Enfermedades Moleculares Madrid	2
Mioadenilato desaminasa deficiency, mutación Q12X	H. 12 de Octubre	36
Propionic acidemia	Centro de Diagnóstico de Enfermedades Moleculares Madrid	25
Propionic acidemia (Genes PCCA and PCCB)	Centro de Diagnóstico de Enfermedades Moleculares Madrid	25
INMUNODEFICIENCIES		
Agammaglobulinemia autosomic (μ chain)	H. La Paz. Madrid	4 families
Agammaglobulinemia X-linked	H. La Paz. Madrid	54 families
Apoptosis deficiency (APO 1)	H. La Paz. Madrid	3 families
Combined Inmunodeficiency X- linked (cad γR-IL2)	H. La Paz. Madrid	5 families
Hyper IgM syndrome	H. La Paz. Madrid	6 families
RAG 1, RAG 2. Combined autosomic Inmunodeficiency	H. La Paz. Madrid	5 families
MITOCHONDIAL DISEASES		
Leigh, MERRF, MELAS, Pearson, Kerans-Sayre	H. 12 de Octubre	215
OTHER CONDITIONS		
Achondroplasia/Hipocondroplasia (FGFR3)	H. Gregorio Marañón Madrid	50
	H. La Paz. Madrid	44
	F. Jiménez Díaz	20
Azoospermia	F. Jiménez Díaz	40
Coroideremia	F. Jiménez Díaz	10
Cystic fibrosis	H. La Paz. Madrid	93
	H. Ramón y Cajal	53
	F. Jiménez Díaz	50
	H. Gregorio Marañón Madrid	N/S
Deafness, linked to Conexina 26 gene	H. Ramón y Cajal	500
	F. Jiménez Díaz	5

Deafness, mitochondrial	H. Ramón y Cajal	600
Deafness, mitochondrial, mutación A1555G	H. 12 de Octubre	36
Deafness mitochondrial (due to ototoxics)		4
Deafness, sporadic (otoferlina gene)	H. Ramón y Cajal	300
Di George syndrome	F. Jiménez Díaz	N/S
Factor V deficiency	F. Jiménez Díaz	35
	H. Gregorio Marañón Madrid	N/S
GH deficiency	H. La Paz. Madrid	8
Hemochromatosis	F. Jiménez Díaz	207
	H. La Paz. Madrid	162
Mediterranean familiar fever	F. Jiménez Díaz	8
	F. Jiménez Díaz	20
Polimorfisms Metabolism Folato: MTHFR 677C→T	Centro de Diagnóstico de Enfermedades Moleculares Madrid	22
Polimorfisms Metabolism Folato: MTRR 66 G→A	Centro de Diagnóstico de Enfermedades Moleculares Madrid	22
Polycystic Kidney Disease I (PKD1)	H. Ramón y Cajal	37
Porphyria Acute Intermitent (AIP)	H. La Paz. Madrid	29
Porphyria, congenital erythropoietic	H. La Paz. Madrid	N/S
Protrombine gene	F. Jiménez Díaz	35
	H. Gregorio Marañón Madrid	N/S
Retinitis pigmentosa	F. Jiménez Díaz	150
Retinitis pigmentosa, autosomic dominant (genes RHO y RDS)	F. Jiménez Díaz	25
Retinitis pigmentosa X-linked	F. Jiménez Díaz	50
Retinoschisis	F. Jiménez Díaz	30
SOS gene linked conditions	H. La Paz. Madrid	85
SRY gene	H. La Paz. Madrid	19
	F. Jiménez Díaz	10
	H. Gregorio Marañón Madrid	5
Wilson disease (ATP7B)	H. La Paz. Madrid	N/S

MURCIA

Population: 1.190.378 inhabitants.

There is only 1 centre, a public hospital, in which genetic tests are performed. The number of conditions that can be tested in that centres is 12.

NEUROLOGIC CONDITIONS	**Centres**	**N. test 2001**
Alzheimer disease(APOE)	Centro de Bioquímica y Genética Clínica	25
Angelman syndrome	Centro de Bioquímica y Genética Clínica	5
Duchenne - Becker muscular dystrophy	Centro de Bioquímica y Genética Clínica	25
Fragile-X syndrome	Centro de Bioquímica y Genética Clínica	250
Myotonic dystrophy Steinert	Centro de Bioquímica y Genética Clínica	35
Prader Willi syndrome	Centro de Bioquímica y Genética Clínica	15
NEOPLASIC CONDITIONS		
MEN1	Centro de Bioquímica y Genética Clínica	25
Multiple Endocrine Neoplasia MEN2A	Centro de Bioquímica y Genética Clínica	25
Lynch syndrome (HNPCC)	Centro de Bioquímica y Genética Clínica	15
OTHER CONDITIONS		
Achondroplasia/Hipocondroplasia (FGFR3)	Centro de Bioquímica y Genética Clínica	2
Azoospermia	Centro de Bioquímica y Genética Clínica	10
Cystic fibrosis	Centro de Bioquímica y Genética Clínica	50

NAVARRA

Population: 556.263 inhabitants.

There are 2 centres, 1 public hospital and 1 private hospital, in which genetic tests are performed.

The number of conditions that can be tested in those centres is 44.

NEUROLOGIC CONDITIONS	Centres	N. test 2001
Alzheimer disease(Presenilina 1, APP, APOE)	H Virgen del Camino Clínica Universitaria Pamplona	125 15
Angelman syndrome	H Virgen del Camino	10-12
Charcot-Marie-Tooth neuropathy	Clínica Universitaria Pamplona	8
Dentatorubral-pallidoluysian atrophy (DRPLA)	Clínica Universitaria Pamplona	3
Dominant ataxias (ADCAS) SCA1; SCA2.(Machado) SCA3; SCA6; SCA7; SCA8; SCA9, SCA10; SCA12	Clínica Universitaria Pamplona	10
Friedrich ataxia	Clínica Universitaria Pamplona	3
Fronto-temporal dementia	Clínica Universitaria Pamplona	6
Hirschsprung disease	Clínica Universitaria Pamplona	18
Huntington disease	H Virgen del Camino	50-60
Kennedy disease (AR, exón 1)	Clínica Universitaria Pamplona	3
Leber optic atrophy	Clínica Universitaria Pamplona	5
Mental retardation FRAXE type linked (FMR2)	H Virgen del Camino	60
Myotonic dystrophy Steinert	H Virgen del Camino	25
Parkinson (Parkin, a-sinucleina)	Clínica Universitaria Pamplona	8
Prader Willi syndrome	H Virgen del Camino	10-12
Torsion dystonia (DYT1)	Clínica Universitaria Pamplona	15
X-Frágile syndrome (FMR1)	H Virgen del Camino	60
HEMATOLOGIC CONDITIONS		
Leukemias	H Virgen del Camino	40-60
Metilen TetraHidrofolato Reductasa (MTHFR)	H Virgen del Camino	250
Thrombophilia (Mut G1691A factor V) (Mut G20210A protrombina)	H Virgen del Camino	250
NEOPLASIC CONDITIONS		
Adenomatous polyposis of the colon (APC)	Clínica Universitaria Pamplona H Virgen del Camino	36 25-30
Adenomatous polyposis of the colon (Gen P53, Gen kRAS, Gen hMHS2, Gen hMLH1, Gen hPMS1,Gen hPMS2)	Clínica Universitaria Pamplona	37
Breast cancer (BCRA1 y BCRA2)	Clínica Universitaria Pamplona	50
CK-19	Clínica Universitaria Pamplona	178
Familiar feochromocitome	Clínica Universitaria Pamplona	3
Familiar melanoma (pl5, p19,CDK4)	Clínica Universitaria Pamplona	12
Familiar melanoma (pl6)	Clínica Universitaria Pamplona	12
GST mu (colon, bladder)	Clínica Universitaria Pamplona	59
K-RAS (CODON 12, 13) (lung, colon, ovary, larynx)	Clínica Universitaria Pamplona	185
Li-Fraumeni disease (p53)	Clínica Universitaria Pamplona	9
Multiple Endocrine Neoplasia type 1 (MEN1)	Clínica Universitaria Pamplona	7
Multiple Endocrine Neoplasia type II A (MEN2A)	Clínica Universitaria Pamplona	19
p53 EXONES 4,5,6,7,8 (Lung, breast, ovary, Endomet.)	Clínica Universitaria Pamplona	289
Von Hippel Lindau disease	Clínica Universitaria Pamplona	12
PSA (prostate cancer)	Clínica Universitaria Pamplona	150
OTHER CONDITIONS		
Azoospermia	H Virgen del Camino	5-10
Celiac disease	H Virgen del Camino	250

Cystic fibrosis	H Virgen del Camino	40-45
Deafness, linked to Conexina 26 gene	Clínica Universitaria Pamplona	156
Di George syndrome	H Virgen del Camino	10-12
Hemochromatosis	H Virgen del Camino	150-175
Morris Syndrome, de Reifenstein Syndrome (AR exones 2-8)	Clínica Universitaria Pamplona	5
SRY gene	H Virgen del Camino	5-10
Wilson disease (ATP7B)	Clínica Universitaria Pamplona	12

PAÍS VASCO

Population: 2.101.478 inhabitants in the year 2001.

There are 4 centres in which genetic tests are performed, of them 3 are public hospitals and and 1 private.

The number of conditions that can be tested in those centres is 29.

NEUROLOGIC CONDITIONS	**Centres**	**N. test 2001**
Distrofia facio-escapulo-humeral	H. Donostia. San Sebastián	100
Fragile-X syndrome	H. Basurto Bilbao	268
Hirschsprung disease	H. Cruces Baracaldo	10
Mental retardation FRAXE type linked (FMR2)	H. Basurto Bilbao	154
Myotonic dystrophy Steinert	H. Donostia. San Sebastián H. Cruces Baracaldo H. Basurto Bilbao	44 25 15
Myotonic distrophy of waists (LGMD) type 2A (Calpainopatía,LGMD2A)	H. Donostia. San Sebastián	74
Parkinson (Parkin, a-sinucleina)	H. Donostia. San Sebastián	26
METABOLIC CONDITIONS		
17-beta-hidroxiesteroide-deshidrogenasa deficiency	H. de Cruces Baracaldo	2
Diabetes type MODY. (GK, HNF1)	H. de Cruces Baracaldo	25
NEOPLASIC CONDITIONS		
Familiar feochromocitome	H. Cruces Baracaldo	3
K-RAS (CODON 12, 13)(lung, colon, ovary, larynx)	H. Cruces Baracaldo	50
MEN1	H. Cruces Baracaldo	40
Multiple Endocrine Neoplasia MEN2A	H. Cruces Baracaldo	5
Multiple Endocrine Neoplasia MEN2B	H. Cruces Baracaldo	5
p53 EXONES 4,5,6,7,8 (Lung, breast, ovary, endometrio)	H. Cruces Baracaldo	50
Thyroid cancer	H. Cruces Baracaldo	15
OTHER CONDITIONS		
Androgenic Resistances: Morris syndrome, Reifenstein syndrome (AR exones 2-8)	H. Cruces Baracaldo	5
Celiac disease (HLA markers)	Policlinica Gipuzkoa. San Sebastián	25
Cystic fibrosis	H. Cruces Baracaldo	32
Espondiloartropathy (genotyping b27)	Policlinica Gipuzkoa. San Sebastián	N/S
Factor V deficiency	H. Cruces Baracaldo	600
Hemochromatosis	H. Cruces Baracaldo	968
Hemochromatosis(Mutations C282Y, H63D, S65C)	Policlinica Gipuzkoa. San Sebastián	15
Narcolepsia (hla markers)	Policlinica Gipuzkoa. San Sebastián	
Panhipopituitarism	H. Cruces Baracaldo	10
Protrombine gene	H. Cruces Baracaldo	600
Pseudohipoparatiroidism	H. Cruces Baracaldo	10
Reumathoid Artritis (hla markers)	Policlinica Gipuzkoa. San Sebastián	N/S
SRY gene	H. Cruces Baracaldo	5

RIOJA

Population: 270.400 inhabitants.

There is only a public hospital in which genetic tests are performed.

The number of conditions that can be tested in that centre is 9.

METABOLIC CONDITIONS	Centres	N. test 2001
11-beta-hidroxilasa deficiency	H. San Millán. Logroño	N/S
17-alfa-hidroxilasa deficiency	H. San Millán. Logroño	N/S
21-hidroxilasa deficiency	H. San Millán. Logroño	N/S
Alpha-1-antitripsina deficiency	H. San Millán. Logroño	N/S
GH deficiency	H. San Millán. Logroño	N/S
Primary hyperaldosteronism	H. San Millán. Logroño	N/S
OTHER CONDITIONS		
Celiac disease	H. San Millán. Logroño	N/S
GH deficiency	H. San Millán. Logroño	N/S
Hemochromatosis	H. San Millán. Logroño	110

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